Les notions de santé et de maladie à l'ère du numérique

Analyse politique et recommandations du Conseil suisse de la science CSS
Policy analysis and recommendations by the Swiss Science Council SSC

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Health and disease in the era of big data
Concept analysis by Prof. Dominic Murphy, University of Sydney

Schweizerische Eidgenossenschaft
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Schweizerischer Wissenschaftsrat
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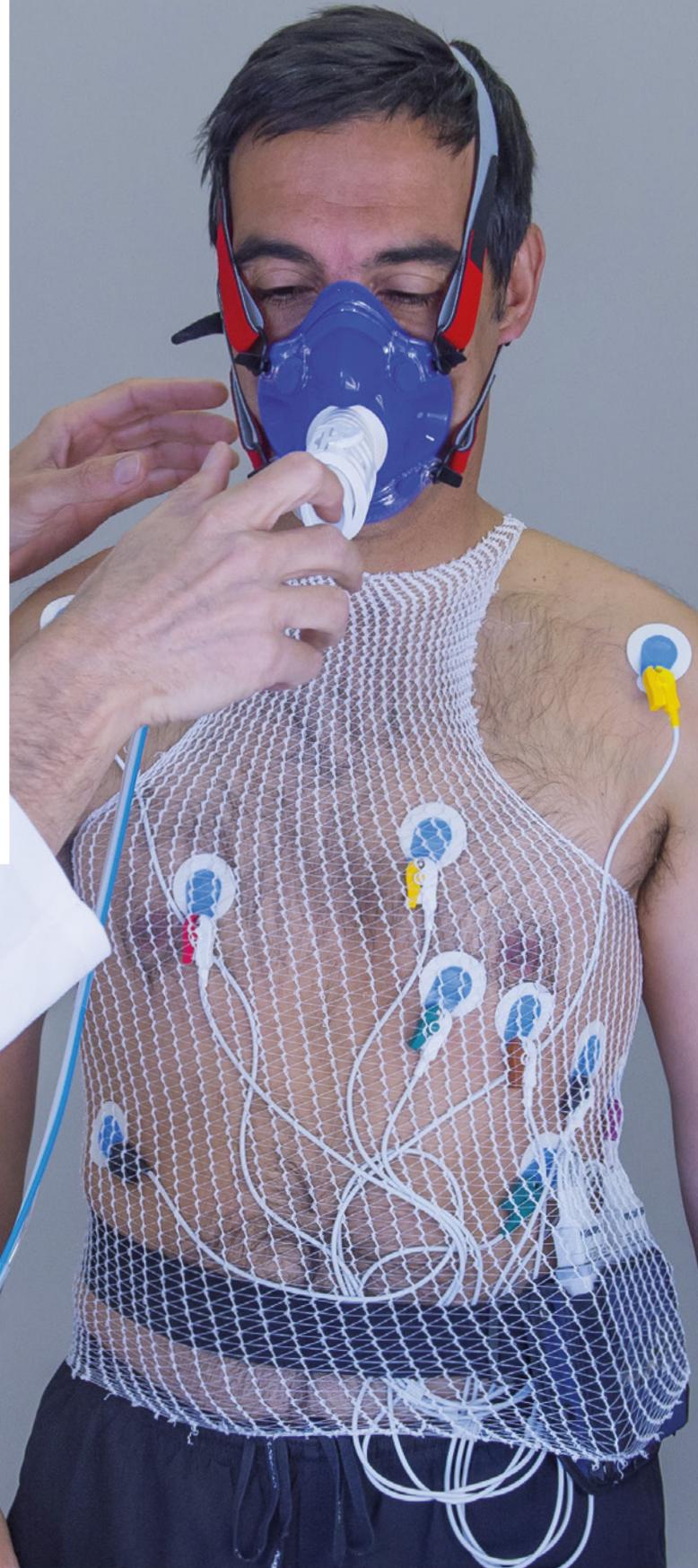
«Nous sommes dans une phase d'incertitude. Nous ne savons pas où nous allons mais nous y allons. Pour nous rassurer nous cherchons de l'information, alors qu'il faudrait chercher du sens, construire un vrai projet appuyé sur une vision commune de la santé et de la maladie. Dès que les gens comprendront le sens du projet sanitaire, ils seront prêts à mettre leur information à disposition. Dès que les médecins verront l'intérêt scientifique et pratique des données, ils participeront.»

Charles Kleiber, Berne, 17 janvier 2019
Atelier de l'Académie suisse des sciences médicales

Le Conseil suisse de la science

Le Conseil suisse de la science CSS est l'organe consultatif du Conseil fédéral pour les questions relevant de la politique de la science, des hautes écoles, de la recherche et de l'innovation. Le but de son travail est l'amélioration constante des conditions-cadre de l'espace suisse de la formation, de la recherche et de l'innovation en vue de son développement optimal. En tant qu'organe consultatif indépendant, le CSS prend position dans une perspective à long terme sur le système suisse de formation, de recherche et d'innovation.

Der Schweizerische Wissenschaftsrat

Der Schweizerische Wissenschaftsrat SWR berät den Bund in allen Fragen der Wissenschafts-, Hochschul-, Forschungs- und Innovationspolitik. Ziel seiner Arbeit ist die kontinuierliche Optimierung der Rahmenbedingungen für die gedeihliche Entwicklung der Schweizer Bildungs-, Forschungs- und Innovationslandschaft. Als unabhängiges Beratungsorgan des Bundesrates nimmt der SWR eine Langzeitperspektive auf das gesamte BFI-System ein.

Il Consiglio svizzero della scienza

Il Consiglio svizzero della scienza CSS è l'organo consultivo del Consiglio federale per le questioni riguardanti la politica in materia di scienza, scuole universitarie, ricerca e innovazione. L'obiettivo del suo lavoro è migliorare le condizioni quadro per lo spazio svizzero della formazione, della ricerca e dell'innovazione affinché possa svilupparsi in modo armonioso. In qualità di organo consultivo indipendente del Consiglio federale il CSS guarda al sistema svizzero della formazione, della ricerca e dell'innovazione in una prospettiva globale e a lungo termine.

The Swiss Science Council

The Swiss Science Council SSC is the advisory body to the Federal Council for issues related to science, higher education, research and innovation policy. The goal of the SSC, in conformity with its role as an independent consultative body, is to promote the framework for the successful development of the Swiss higher education, research and innovation system. As an independent advisory body to the Federal Council, the SSC pursues the Swiss higher education, research and innovation landscape from a long-term perspective.

Préface du CSS

Dans le cadre de son programme de travail 2016–2019, le CSS analyse dans quelle mesure la numérisation et les approches scientifiques faisant appel aux grands ensembles de données influent sur les pratiques dans le système de santé. D'une part, le Conseil a chargé la Prof. Sabina Leonelli, de l'Université d'Exeter, de réaliser une étude intitulée «La production de connaissances biomédicales à l'ère du Big Data», s'interrogeant sur la fonction des données dans la démarche scientifique et explorant les opportunités et défis liés à leur gestion et à leur interprétation. Ce travail a donné lieu à une publication parue fin 2017[1]. D'autre part, une analyse des concepts de santé et de maladie a été confiée au Prof. Dominic Murphy de l'Université de Sydney. Son étude présente les principales positions philosophiques sur la définition de la santé et de la maladie, et s'interroge sur l'impact de l'utilisation de grands ensembles de données pour ces définitions. Cette analyse est publiée en deuxième partie du présent rapport. Dans la première partie (disponible en français et en anglais), le CSS présente ses considérations et recommandations en matière de politique de santé et de politique de formation, recherche et innovation, développées sur la base des études exploratoires et de divers échanges menés avec une large palette d'experts. Par ailleurs, de concert avec l'Académie suisse des sciences médicales, le Conseil publie séparément un compte rendu de l'atelier commun organisé en avril 2018 et consacré à la question: «Patient, médecin, big data. Qui a le pouvoir de définition?».[2]

Vorwort des SWR

Im Rahmen seines Arbeitsprogramms 2016–2019 analysiert der SWR, inwiefern die Digitalisierung und datengestützte wissenschaftliche Ansätze die Praktiken im Gesundheitswesen beeinflussen. Zum einen hat der Rat Prof. Sabina Leonelli von der Universität Exeter beauftragt, die Studie «Wissensproduktion in der Biomedizin im Zeitalter von Big Data»[3] zu erarbeiten. Diese Ende 2017 veröffentlichte Analyse untersucht die Funktion von Daten im wissenschaftlichen Prozess und erläutert Chancen und Herausforderungen im Zusammenhang mit der Datenverwaltung und -interpretation. Zum anderen wurde Prof. Dominic Murphy von der Universität Sydney mit einer Analyse der Konzepte von Gesundheit und Krankheit beauftragt. Seine Studie beschreibt die wichtigsten philosophischen Positionen zur Definition von Gesundheit und Krankheit und befasst sich mit der Frage, wie sich die Arbeit mit grossen Datensätzen auf die Definitionen auswirken könnte. Diese Analyse wird im zweiten Teil des vorliegenden Berichts veröffentlicht. Im ersten Teil (verfügbar in Französisch und Englisch, deutsche Zusammenfassung siehe S. 7–8) stellt der SWR seine Überlegungen und Empfehlungen zur Gesundheitspolitik sowie zur Politik im Bereich Bildung, Forschung und Innovation vor. Diese stützen sich auf die explorativen Studien und den Austausch mit verschiedensten Fachleuten. Zudem veröffentlicht der Rat zusammen mit der Schweizerischen Akademie der Medizinischen Wissenschaften einen separaten Bericht zum gemeinsamen Workshop vom April 2018 zum Thema «Patient, Arzt, Big Data – wer hat die Definitionsmacht?».[4]

1   S. Leonelli (2017). La production de connaissances biomédicales à l'ère du Big Data. Analyse réalisée sur mandat du Conseil suisse de la science et de l'innovation CSSI. Étude exploratoire 2/2017. Rapport en anglais.

2   ASSM & CSS (2019). Patient, médecin, big data. Qui a le pouvoir de définition? Swiss Academies Communications. Vol. 14, No 3, 2019. Rapport disponible en français, allemand et anglais.

3   S. Leonelli (2017). Wissensproduktion in der Biomedizin im Zeitalter von Big Data. Analyse im Auftrag des Schweizerischen Wissenschafts- und Innovationsrates SWIR. Explorative Studie 2/2017. Bericht in englischer Sprache.

4   SAMW & SWR (2019). Patient, Arzt, Big Data – wer hat die Definitionsmacht? Swiss Academies Communications. Vol. 14, No 3, 2019. Bericht in englischer, französischer und deutscher Sprache.

Prefazione del CSS

Nell'ambito del suo Programma di lavoro 2016–2019 il CSS analizza in che misura la digitalizzazione e gli approcci scientifici che ricorrono a enormi quantità di dati influenzano le pratiche del sistema sanitario. Da un lato il CSS ha invitato la professoressa Sabina Leonelli dell'Università di Exeter a condurre uno studio sulla genesi del sapere biomedico nell'era dei *big data*. Pubblicata alla fine del 2017, quest'indagine esamina la funzione di dati nel processo scientifico ed esplora le opportunità e le sfide legate alla loro gestione e interpretazione[5]. Dall'altro lato, un'analisi delle nozioni di salute e malattia è stata commissionata al professor Dominic Murphy dell'Università di Sydney. Il suo studio presenta le principali prospettive filosofiche relative a definizioni di salute e malattia e si interroga su come queste definizioni si stanno sviluppando sotto l'impatto dei *big data*. Quest'analisi è pubblicata nella seconda parte del presente rapporto. Nella prima (in francese e inglese, con riassunto in italiano alle pagine 9-10), il CSS presenta le sue riflessioni e raccomandazioni in materia di politica sanitaria, di educazione, ricerca e innovazione, sviluppate sulla base di studi esplorativi e scambi con una vasta gamma di esperti. Inoltre, in collaborazione con l'Accademia svizzera delle scienze mediche, il CSS pubblica separatamente un rapporto sul workshop collettivo tenutosi in aprile 2018 dal tema: «Pazienti, medici e *big data*: chi ha il potere decisionale?».[6]

Preface by the SSC

As part of its Working Programme 2016–2019, the SSC investigates the extent to which digitalisation and data-driven scientific approaches influence practices in the health care system. On the one hand, the Council invited Prof. Sabina Leonelli of the University of Exeter, to conduct the study "Biomedical knowledge production in the age of big data"[7], examining the function of data in the scientific process and exploring opportunities and challenges related to data management and interpretation. This work resulted in a publication at the end of 2017. On the other hand, an analysis of the concepts of health and disease was commissioned to Prof. Dominic Murphy of the University of Sydney. His study presents the main philosophical positions on defining health and disease and asks how the increased use of data-driven approaches might influence such definitions. This analysis is published in the second part of the present report. In the first part (available in French and English), the SSC presents its considerations and recommendations on health policy as well as education, research and innovation policy, developed on the basis of exploratory studies and various exchanges conducted with a wide range of experts. In addition, the Council, in conjunction with the Swiss Academy of Medical Sciences, publishes a separate report on the joint workshop held in April 2018 on the subject: "Patient, doctor, big data. Who has the power of definition?".[8]

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[5]   S. Leonelli (2017). *Produzione di conoscenze biomediche nell'era dei big data. Analisi svolta su incarico del Consiglio svizzero della scienza e dell'innovazione CSSI. Studio esplorativo 2/2017 in inglese.*

[6]   ASSM & CSS (2019). «Patient, médecin, big data. Qui a le pouvoir de définition?», Swiss Academies Communications. Vol. 14, No 3, 2019. (in francese, tedesco e inglese).

[7]   S. Leonelli (2017). *Biomedical knowledge production in the age of big data. Analysis conducted on behalf of the Swiss Science and Innovation Council SSIC. Exploratory study 2/2017, in English.*

[8]   SAMS & SSC (2019). *Patient, doctor, big data. Who has the power of definition? Swiss Academies Communications. Vol. 14, No 3, 2019. Report available in English, French and German.*

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Résumé

Le Conseil suisse de la science (CSS) a étudié l'impact de la numérisation sur la pratique de la médecine. Certaines questions fondamentales persistent, en particulier le fait que les concepts de santé et de maladie ne peuvent s'appuyer uniquement sur des résultats empiriques, car ils sont fortement influencés par le contexte et les aspirations sociales. C'est pourquoi il semble préférable de parler de notions multiples plutôt que d'un seul concept de santé. À l'ère du numérique, quatre perspectives sur la santé et la médecine paraissent particulièrement pertinentes: la première est celle de la science, où la santé est définie comme un potentiel ou un risque calculable; la deuxième est celle de l'éthique, et la traduction de valeurs en priorités et en choix; la troisième est celle de l'économie, et le débat persistant sur l'augmentation des coûts; et, enfin, la quatrième consiste à évaluer comment la pratique professionnelle de la médecine et des soins de santé peut évoluer afin de tirer profit des nouveaux instruments et méthodes.

En se basant sur des études exploratoires et des échanges avec différents experts, le CSS estime que les approches fondées sur des données seront très profitables à la pratique de la médecine, à condition que les présupposés scientifiques sur lesquels les données reposent soient pris en compte, au même titre que les normes sociales et les valeurs individuelles intervenant dans toute application des soins de santé. L'intelligence artificielle et l'apprentissage automatique peuvent faire progresser la médecine si leur résultat est placé dans le bon contexte par des utilisateurs expérimentés et responsables.

Pour contribuer à relever ces défis, le CSS a formulé une série de recommandations relatives à la politique de la santé et de la science. Les recommandations s'adressent essentiellement au Conseil fédéral, au Secrétariat d'État à la formation, à la recherche et à l'innovation (SEFRI), à l'Office fédéral de la santé publique (OFSP) ainsi qu'aux autorités cantonales responsables des départements de la santé et de l'éducation. Les recommandations ne portent pas sur des questions spécifiques en particulier, mais ont pour objectif de sensibiliser à la nécessité d'une politique cohérente en matière d'utilisation du *big data* dans le système des soins de santé. Le chapitre 5.2 de la présente analyse politique détaille le raisonnement qui soustend chaque recommandation.

Promouvoir une compréhension globale de la santé

Au Conseil fédéral, le CSS recommande
1. de considérer la formation de base, les programmes sociaux et les questions environnementales comme des investissements prioritaires en tant que mesures de promotion de la santé à part entière.

À l'OFSP et au SEFRI, le CSS recommande
2. d'assurer le soutien durable de la recherche sur les services de santé;

3. d'encourager un débat public continu impliquant toutes les composantes de la société pour déterminer les attentes en matière de soins de santé et de qualité de vie.

Développer des standards de données favorables à la santé

À l'OFSP, le CSS recommande
4. de réduire le biais de publication dans la littérature clinique en assurant l'accès aux résultats des essais cliniques;
5. d'augmenter les compétences et de définir les critères de validation pour les logiciels médicaux.

Au SEFRI et aux autorités cantonales, le CSS recommande
6. de soutenir les services essentiels tels que l'annotation de données et la conservation de bases de données.

À l'OFSP et au SEFRI, le CSS recommande
7. de garantir la prise en compte des implications sociales liées au choix des données et à leur spécification.

Préciser les objectifs de l'initiative Swiss Personalized Health Network (SPHN)

Au SEFRI, à l'OFSP et aux autorités cantonales, le CSS recommande
8. de créer un centre national pour l'informatique médicale et l'analyse des données médicales, qui définira des normes et orientera l'ensemble des institutions médicales suisses dans le contexte de l'initiative SPHN.

Au SEFRI, le CSS recommande
9. de définir la valeur ajoutée attendue des projets scientifiques, de la collaboration institutionnelle et du partage de pratiques soutenus par l'initiative SPHN, et d'aider à positionner le SPHN au niveau international.

Repenser la formation médicale

À l'OFSP, au SEFRI et aux autorités cantonales, le Conseil recommande
10. de définir un socle de compétences commun pour le bachelor en médecine et de proposer un petit nombre de masters différenciés en médecine;
11. de développer de nouveaux programmes de formation dans des disciplines voisines, au-delà du cursus de formation médicale.

Zusammenfassung

Der Schweizerische Wissenschaftsrat (SWR) hat den Einfluss der Digitalisierung auf die medizinische Praxis untersucht. Gewisse grundlegende Probleme bleiben bestehen; so insbesondere, dass das Verständnis von Gesundheit und Krankheit nie ausschliesslich auf empirischen Fakten gründen kann, da es stark vom Kontext und gesellschaftlichen Erwartungen beeinflusst wird. Deshalb scheint es sinnvoller, im Zusammenhang mit Gesundheit von mehreren Auffassungen anstatt von einem einzigen Konzept zu sprechen. Im digitalen Zeitalter scheinen vier Perspektiven auf Gesundheit und Medizin besonders wichtig: Erstens jene der Wissenschaft, die Gesundheit als kalkulierbares Potenzial oder Risiko begreift; zweitens jene der Ethik, die um die Übersetzung von Werten in Prioritäten und Entscheidungen bemüht ist; drittens jene der Wirtschaft und der anhaltenden Debatte über die Zunahme der Kosten; und schliesslich gilt es auch zu berücksichtigen, wie sich die berufliche Praxis in Medizin und Pflege weiterentwickeln lässt, um die neuen Instrumente und Methoden zu nutzen.

Gestützt auf explorative Studien und den Austausch mit verschiedenen Fachleuten kommt der SWR zur Einschätzung, dass datengestützte Ansätze wichtige Erkenntnisse zur medizinischen Praxis liefern werden. Dies setzt allerdings voraus, dass die wissenschaftlichen Annahmen, auf denen die Daten basieren, die gesellschaftlichen Normen sowie individuelle Werte, die den therapeutischen Interventionen zu Grunde liegen, berücksichtigt werden. Künstliche Intelligenz und maschinelles Lernen können die Medizin voranbringen, sofern die Ergebnisse von erfahrenen, verantwortungsvollen Anwenderinnen und Anwendern in den richtigen Kontext gestellt werden.

Um diese Herausforderungen anzugehen, hat der SWR eine Reihe von Empfehlungen sowohl für die Gesundheits- als auch für die Wissenschaftspolitik formuliert. Sie sind in erster Linie als Unterstützung für den Bundesrat, das Staatssekretariat für Bildung, Forschung und Innovation (SBFI), das Bundesamt für Gesundheit (BAG) sowie die kantonalen Gesundheits- und Bildungsbehörden gedacht. Die Empfehlungen gehen nicht detailliert auf einzelne Probleme ein, sondern sollen das Bewusstsein dafür schärfen, dass es im Gesundheitswesen eine kohärente Politik für die Nutzung von ‹Big Data› braucht. Die Begründungen für die einzelnen Empfehlungen sind in Kapitel 5.2 der politischen Analyse zu finden.

Zur Förderung eines umfassenden Verständnisses von Gesundheit

empfiehlt der SWR dem Bundesrat,
1. Investitionen in die Grundbildung, in soziale Programme und den Umweltschutz als wesentliche Bestandteile gesundheitsfördernder Massnahmen prioritär zu behandeln;

empfiehlt der SWR dem BAG und dem SBFI,
2. eine langfristige Unterstützung der Versorgungsforschung sicherzustellen;

3. eine kontinuierliche Debatte anzustossen, die alle Teile der Bevölkerung einbezieht, um gesellschaftliche Erwartungen in Bezug auf Gesundheitsversorgung und Lebensqualität in Erfahrung zu bringen.

Zur Entwicklung gesundheitsfördernder Datenstandards

empfiehlt der SWR dem BAG,
4. den Publikationsbias in der klinischen Literatur zu reduzieren, indem der Zugang zu den Ergebnissen klinischer Studien sichergestellt wird;
5. Kompetenzen auszubauen und Validierungskriterien für medizinische Software zu definieren;

empfiehlt der SWR dem SBFI und den kantonalen Behörden,
6. zentrale Dienstleistungen wie die Datenannotation und die Kuration von Datenbanken zu unterstützen;

empfiehlt der SWR dem BAG und dem SBFI,
7. zu gewährleisten, dass die gesellschaftlichen Auswirkungen der Datenauswahl und -spezifikation berücksichtigt werden.

Zur Präzisierung der Ziele des Swiss Personalized Health Network (SPHN)

empfiehlt der SWR dem SBFI, dem BAG und den Kantonen,
8. ein nationales Zentrum für medizinische Informatik und medizinische Datenanalyse zu schaffen, das Standards festlegt und sämtlichen medizinischen Einrichtungen der Schweiz im Kontext der SPHN-Initiative als Orientierungshilfe dient;

empfiehlt der SWR dem SBFI,
9. den erwarteten Mehrwert von wissenschaftlichen Projekten, institutionellen Kooperationen und gemeinsamen Praktiken, die durch die SPHN-Initiative gefördert werden, zu bestimmen und die internationale Positionierung des SPHN zu unterstützen.

Um ein Umdenken in der medizinischen Ausbildung zu bewirken,

empfiehlt der SWR dem BAG, dem SBFI und den Kantonen,
10. Kernkompetenzen für den Bachelor in Medizin zu definieren und eine kleine Anzahl differenzierter Master-Studiengänge in Medizin anzubieten;
11. über das Medizinstudium hinaus neue Curricula an den Schnittstellen zu anderen Fächern zu entwickeln.

Riassunto

Il Consiglio svizzero della scienza (CSS) ha studiato l'impatto della digitalizzazione sulla medicina. Alcuni aspetti fondamentali rimangono invariati, in particolare il fatto che le nozioni di salute e malattia – fortemente influenzate dal contesto e dalle aspettative sociali – non potranno mai basarsi esclusivamente su dati empirici. È quindi più utile parlare di «nozioni multiple», anziché limitarsi a un singolo concetto di salute. Nell'era digitale quattro prospettive sembrano particolarmente importanti: al primo posto c'è la scienza, che provvede a classificare la salute in termini di potenzialità e rischi calcolabili; al secondo l'etica, che traduce valori in priorità e scelte; al terzo l'economia, alle prese con la problematica di lungo termine del costante aumento dei costi e, per finire, le professioni sanitarie stesse e la questione di come svilupparle e far sì che possano mettere a frutto nuovi strumenti e metodi.

Basandosi su studi esplorativi e scambi con esperti, il CSS concorda sul fatto che gli approcci basati sui big data consentiranno di generare conoscenze utili per l'esercizio delle professioni mediche, a condizione, però, che prendano in considerazione le premesse scientifiche sulle quali queste grandi raccolte di dati si basano nonché le norme sociali e i valori individuali che confluiscono in qualsiasi applicazione sanitaria. L'intelligenza artificiale e l'apprendimento automatico possono senz'altro far progredire la medicina. I risultati che producono vanno però collocati nel giusto contesto da utenti esperti e responsabili.

Per contribuire ad affrontare queste sfide, il CSS ha formulato una serie di raccomandazioni in materia di politica sanitaria e scientifica. L'obiettivo principale è dar man forte al Consiglio federale, alla Segreteria di Stato per la formazione, la ricerca e l'innovazione (SEFRI), all'Ufficio federale della sanità pubblica (UFSP) e ai dipartimenti cantonali della sanità e dell'educazione. Le raccomandazioni non affrontano questioni specifiche, ma sono intese a sensibilizzare sulla necessità di adottare una politica coerente di utilizzo dei «big data» nel sistema sanitario. Gli argomenti sottesi alle singole raccomandazioni si trovano alla sezione 5.2 della presente analisi politica.

Promuovere un'ampia comprensione del concetto di salute

Al Consiglio federale il CSS raccomanda di
1. dare la priorità agli investimenti nell'educazione di base, nei programmi sociali e nelle questioni ambientali come parti integranti delle misure di promozione della salute.

All'UFSP e alla SEFRI il CSS raccomanda di
2. garantire a lungo termine il sostegno alla ricerca sui servizi sanitari, e

3. promuovere un dialogo continuo che coinvolga tutti i ceti sociali e sia finalizzato a conoscere le aspettative pubbliche in materia di sanità e qualità di vita.

Sviluppare standard di dati ritagliati sul sistema sanitario

All'UFSP il CSS raccomanda di
4. ridurre le distorsioni nella letteratura scientifica («publication bias») garantendo l'accesso ai risultati degli studi clinici, e
5. aumentare la competenza e definire criteri di valutazione per i software sanitari.

Alla SEFRI e alle autorità cantonali il CSS raccomanda di
6. sostenere servizi essenziali come l'annotazione di dati e la cura dei database.

All'UFSP e alla SEFRI il CSS raccomanda di
7. sincerarsi che siano prese in considerazione le implicazioni sociali derivanti dalla scelta e dalla specificazione dei dati.

Specificare gli obiettivi dell'iniziativa Swiss Personalized Health Network (SPHN)

Alla SEFRI, all'UFSP e alle autorità cantonali il CSS raccomanda di
8. istituire un centro nazionale per l'informatica medica e l'analisi dei dati medici che dovrà definire gli standard e fornire un quadro orientativo all'intero sistema sanitario svizzero nell'ambito dell'iniziativa SPHN.

Alla SEFRI il CSS raccomanda di
9. definire il valore aggiunto previsto dei progetti scientifici, della collaborazione istituzionale e delle pratiche di condivisione promosse dall'iniziativa SPHN e contribuire a posizionare quest'ultima a livello internazionale.

Ripensare la formazione medica

All'UFSP, alla SEFRI e alle autorità cantonali il CSS raccomanda di

10. definire un catalogo condiviso di competenze da coprire con gli studi di bachelor in medicina e offrire, sempre in medicina, un esiguo numero di master differenziati, e
11. sviluppare nuovi programmi di studio interdisciplinari che sconfinino in ambiti affini.

Executive summary

The Swiss Science Council (SSC) has examined the impact of digitalisation on the practice of medicine. Certain fundamental issues persist, most prominently, that the concept of health and disease can never be grounded entirely on empirical fact, as it is strongly influenced by context and social expectations. Therefore, it appears more appropriate to speak of multiple notions, rather than of a single concept of health. Four perspectives on health and medicine seem particularly relevant in the digital age: first science, or the framing of health as a computable potential or risk; second ethics, and the translation of values into priorities and choices; third economics, and the long-standing debate on cost increases; and, finally, one must consider how the professional practice of medicine and care can evolve to make use of novel instruments and methods.

Based on exploratory studies and exchanges with various experts, the SSC agrees that data-driven approaches will bring important insights to the practice of medicine, provided the scientific assumptions underlying the data as well as the social norms and individual values that feed into any health care application are taken into account. Artificial intelligence and machine learning can enhance medicine, provided their output is placed in the appropriate context by experienced, responsible users.

To help address these challenges, the SSC has formulated a series of recommendations pertaining to both health and science policy. The recommendations are primarily addressed at the Federal Council, the State Secretariat for Education, Research and Innovation (SERI), the Federal Office on Public Health (FOPH) as well as the cantonal authorities in charge of the health and education departments. The recommendations do not address single issues in detail but are intended to raise awareness of the need for a coherent policy regarding the use of "big data" in the health care system. The rationale for each recommendation is to be found in section 5.2 of the policy analysis.

Promote a comprehensive understanding of health

To the Federal Council, the SSC recommends to
1. prioritise investments in basic education, social programmes and environment issues as an integral part of health-promoting measures.

To the FOPH and the SERI, the SSC recommends to
2. ensure long-term support of health services research;

3. encourage a continuous debate that includes all strata of society, to explore public expectations with respect to health care and quality of life.

Develop data standards conducive to health

To the FOPH, the SSC recommends to
4. reduce publication bias in the clinical literature by ensuring access to the results of clinical trials;
5. increase expertise and define validation criteria for medical software.

To the SERI and the cantonal authorities, the SSC recommends to
6. support critical data services such as annotation and database curation.

To the FOPH and the SERI, the SSC recommends to
7. ensure that the social implications of data choice and specification are taken into account.

Precisely define the goals of the Swiss Personalized Health Network (SPHN)

To the SERI, the FOPH and the cantonal authorities, the SSC recommends to
8. establish a national centre for medical informatics and medical data analysis which will set standards and provide guidance to the entire Swiss medical establishment within the context of the SPHN initiative.

To the SERI, the SSC recommends to
9. define the added value expected for scientific projects, institutional collaboration and sharing practices promoted by the SPHN initiative and to help position the SPHN internationally.

Rethink medical education

To the FOPH, the SERI and the cantonal authorities, the Council recommends to
10. define a common core of competencies to be covered by the Bachelor in Medicine and to propose a small number of differentiated Masters in Medicine;
11. develop new curricula at the boundaries to other disciplines beyond the medical track.

Considérations et recommandations du CSS

Considérations et recommandations du CSS
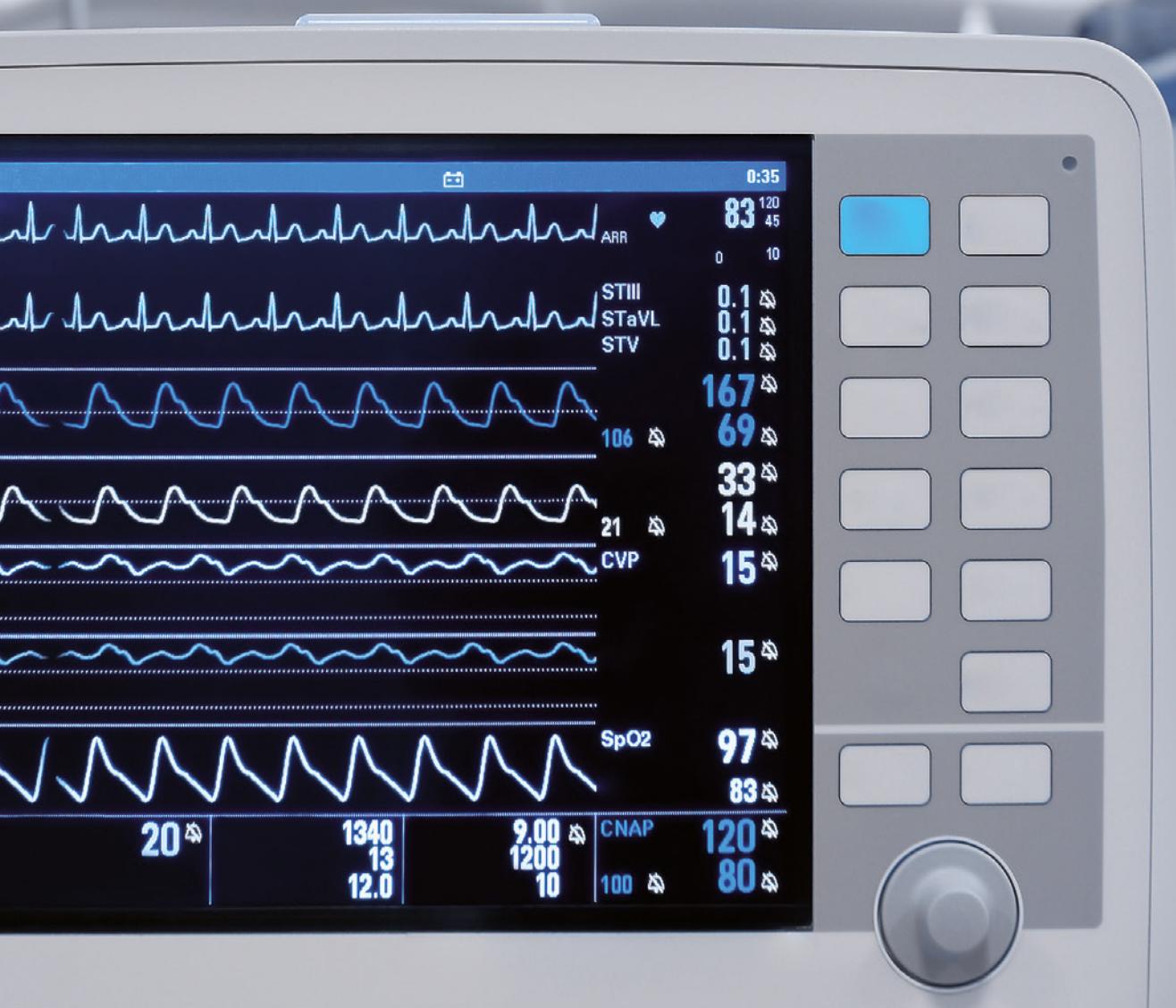
1

Objectif

Dans son programme de travail 2016–2019, le Conseil constate une rapide augmentation de l'accumulation et de la mise à disposition des informations touchant à la santé. La numérisation de la santé a de profondes répercussions pour l'ensemble des acteurs publics et privés. Le Swiss Personalized Health Network (SPHN) et d'autres initiatives nationales ou internationales s'efforcent de réguler et d'intégrer systématiquement ces données dans la pratique de la médecine, tout en tenant compte des pratiques d'autocontrôle qui se multiplient parmi les citoyens[9]. «Le point commun à toutes ces approches est le présupposé selon lequel la ‹norme› en termes de santé est connue de tous et qu'elle peut être définie par des paramètres mesurables, qui peuvent, à leur tour, être consultés et sans cesse précisés grâce à des analyses statistiques.»[10]

Le Conseil souhaite mettre en lumière les défis spécifiques liés à ces nouvelles approches et aux présupposés sur lesquels elles se basent, afin de réfléchir aux conséquences possibles pour la politique publique. Les présupposés sont des affirmations acceptées comme vraies en l'absence de preuve dans le but de constituer un argument. D'un côté, l'allégation selon laquelle le partage des données fait progresser la médecine peut être vérifiée. Par exemple, les jeunes patients atteints de maladies rares peuvent désormais espérer être diagnostiqués rapidement grâce à l'analyse génomique et/ou aux plateformes en ligne où des patients du monde entier partagent leurs symptômes, plutôt que de faire l'expérience d'années d'errance diagnostique aux mains de spécialistes perplexes. De même, la cartographie génétique des mutations d'une centaine d'oncogènes s'est révélée très utile pour définir des thérapies ciblées contre le cancer. D'un autre côté, il est urgent de remettre en question des présupposés courants dans le contexte de la recherche et des soins de santé, par exemple l'idée que «les données parlent d'elles-mêmes», que «le stockage et le transport d'informations numériques ne coûtent presque rien» ou que «la corrélation est un bon substitut au lien de causalité».

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9   Pour ne citer que quelques exemples, on peut souligner la Precision Medicine Initiative aux États-Unis, avec sa cohorte All Of Us, l'initiative phare Médecine personnalisée au Canada et le 100000 Genomes Project au Royaume-Uni. La base de données du Consortium international pour la médecine personnalisée (ICPerMed) accessible à l'adresse https://www.icpermed.eu/ offre une vue plus complète.

10   Programme de travail du CSS 2016–2019, p. 33. Accessible via: https://www.wissenschaftsrat.ch/fr/programme-de-travail.

Prédiction immédiate *(nowcasting)*

2008 a vu le lancement de Google Flu Trends, un service Web pour la surveillance épidémiologique basé sur les requêtes saisies dans le moteur de recherche. Au départ, l'algorithme semblait détecter les épidémies de grippe avec précision et plus rapidement que les centres de contrôle établis, même s'il a entièrement manqué la pandémie non saisonnière A(H1N1) de 2009. Mais en 2015, après avoir surévalué à plusieurs reprises la prévalence des épidémies saisonnières, Google Flu Trends a été arrêté. Les observateurs estiment que les développeurs du service ne parvenaient pas à adapter leur modèle à la dynamique des comportements des utilisateurs, par exemple les phénomènes de surmédiatisation, et aux mises à jour quotidiennes du moteur de recherche Google[11]. Cet exemple met en exergue une série d'enjeux emblématiques de la recherche et du développement faisant appel au *big data*: manque d'accès public aux données primaires et divulgation limitée de l'algorithme, mais aussi corrélation apparemment parfaite grâce au recalibrage, puisque tant l'«instrument de mesure» que l'objet étudié évoluent constamment.

Ce revers ne signifie pas que toute application de *nowcasting* destinée à la surveillance de maladies est vouée à l'échec, surtout si l'application est conçue comme un complément plutôt qu'un substitut à la collecte et à l'analyse traditionnelles de données. Des efforts actuels faisant usage de divers types de sources en ligne pourraient mener à des avancées significatives en matière de santé publique, en particulier pour les pays en voie de développement.

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11   D. Lazer et al. (2014). Big data. The parable of Google Flu: traps in big data analysis. Science. 2014; 343(6176):1203-5.

Cette réflexion n'est pas un manifeste pour ou contre la numérisation dans le secteur de la santé. Étant donné que de plus en plus de données et d'algorithmes sont utilisés, ils ne manqueront pas d'avoir un impact sérieux sur la pratique de la médecine. Pour que leur effet soit bénéfique, des conditions doivent être fixées au sein des institutions de recherche et des établissements de soins en termes de normes de qualité des données et de modes de collaboration. De plus, pour atténuer la tendance à la surmédicalisation déjà manifeste dans les pays à revenus élevés, il convient de réfléchir aux attentes en matière de santé, tant au niveau individuel que sociétal. Le contexte plus large dans lequel les *big data*[12], l'intelligence artificielle (IA)[13] et la médecine personnalisée[14] sont mis en place joue un rôle crucial pour le bien commun. Le présent rapport présente les réflexions du Conseil sur les conditions-cadre nécessaires à l'utilisation de grands volumes de données dans la médecine et la santé en général. Il s'adresse essentiellement aux autorités publiques chargées de guider le développement des systèmes de soins de santé et de formation, qui sont partagés en Suisse entre le niveau fédéral et le niveau cantonal. Ce rapport contribue aussi à enrichir le discours sur la santé et peut intéresser un cercle plus large d'institutions, d'experts et de membres de la société civile, d'autant plus importants dans un système où nombre de compétences ont été déléguées aux acteurs directement concernés.

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12   Pour une définition du *big data* dans les sciences du vivant, voir S. Leonelli (2016). Data-Centric Biology: A Philosophical Study. The University of Chicago Press; ainsi que S. Leonelli (2017). La production de connaissances biomédicales à l'ère du Big Data. Analyse réalisée sur mandat du Conseil de la science et de l'innovation CSSI. Étude exploratoire 2/2017.

13   L'IA est la simulation de l'intelligence humaine par des systèmes informatiques et autres machines. Les applications de l'IA couvrent l'apprentissage automatique qui permet aux systèmes de reconnaître des schémas et de classer des objets tels que des images, des sons ou des textes sans avoir été programmés explicitement pour cette tâche. Par exemple, les systèmes automatisés d'aide aux décisions cliniques analysent les données du patient et aident les professionnels dans leurs diagnostics et leurs prévisions.

14   La médecine personnalisée a pour objectif d'orienter le patient vers un choix ciblé d'options thérapeutiques par le recours à une abondance de données sur son état général et ses caractéristiques individuelles.

2
Méthodes

Pour nourrir sa réflexion, le CSS a créé un groupe de travail interdisciplinaire qui a entrepris une série de discussions et d'échanges, et présenté ses résultats au Conseil à plusieurs occasions. La notion de santé est vaste et multidimensionnelle. Les questions étudiées par le Conseil ne représentent évidemment qu'une sélection parmi tous les aspects et points de vue possibles. Les choix du Conseil restent étroitement liés aux thèmes de l'éducation, de la recherche et de l'innovation, qui sont au cœur de son domaine d'expertise. Étant donné le caractère prospectif des questions et les changements rapides du paysage institutionnel, le Conseil n'a pas pour ambition d'aboutir à une définition définitive de la santé, ni à des prédictions sur l'avenir du système de soins et de la recherche dans le domaine de la santé.

Afin de mieux refléter la multiplicité des points de vue, une approche discursive et coopérative a été retenue en concertation avec des experts et institutions nationaux et internationaux, associant des ateliers, interviews et rapports d'expertises (chapitre 3). Ces contributions d'experts ont été ensuite approfondies et complétées par des analyses de la littérature et des échanges au sein du groupe de travail (chapitre 4). Sur cette base, le CSS présente sa synthèse et ses recommandations (chapitre 5).

3
Contributions d'experts

Le 15 janvier 2018, le groupe de travail du CSS a organisé un séminaire à Zurich sur le potentiel scientifique du *big data*. Les principaux intervenants étaient Ewan Birney, codirecteur du European Bioinformatics Institute of the European Molecular Biology Laboratory (EMBL-EBI) et Stephen Senn, directeur du Centre de Compétences de la Méthodologie et des Statistiques du Luxembourg Institute of Health.

> Pour Ewan Birney, l'immense quantité de données qui sera bientôt générée pour chaque personne va changer la donne. La recherche et les pratiques médicales resteront guidées par différents intérêts: la recherche biomédicale est devenue une entreprise internationale, tandis que les pratiques médicales demeurent intimement liées aux caractéristiques spécifiques des systèmes de santé nationaux. Des instituts supranationaux tels que l'EMBL-EBI définiront des normes et un vocabulaire communs pour la communauté scientifique. Au niveau national, des organisations centralisées devraient être mises en place pour coordonner la conservation et la standardisation des données médicales.

> Pour Stephen Senn, les scientifiques et les régulateurs croient que les données publiées et les méthodes expérimentales sont capables d'indiquer le champ d'application de la médecine personnalisée, alors que c'est rarement le cas. Le grand défi consiste à comprendre la source des variations statistiques. Une hypothèse répandue mais non vérifiée voudrait que les facteurs génétiques soient plus importants que d'autres facteurs de variation tels que l'environnement du patient, les divergences de pratique parmi les médecins ou même les erreurs de mesure. De petits designs expérimentaux bien conçus seraient plus utiles pour répondre à ces questions que de simplement multiplier les mesures sur un nombre de patients toujours plus grand[15].

Le 18 avril 2018, le groupe de travail du CSS a tenu un atelier avec l'Académie suisse des sciences médicales (ASSM). Les intervenants principaux étaient Joachim Buhmann, professeur de sciences informatiques à l'EPFZ, et Werner Bartens, médecin et rédacteur à la *Süddeutsche Zeitung*. Au total, 32 experts y ont participé, et l'ASSM et le CSS ont rendu compte de l'atelier dans une publication commune[16].

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15 Voir aussi: S. Senn (2018). Statistical pitfalls of personalized medicine. Nature. 2018 Nov; 563(7733):619-621.

16 ASSM & CSS (2019). Patient, médecin, big data. Qui a le pouvoir de définition? Swiss Academies Communications. Vol. 14, No 3, 2019.

Pour Joachim Buhmann, l'apprentissage automatique et l'IA auront un profond impact sur la médecine en reprenant de nombreuses tâches et compétences. À long terme, il ne restera probablement aux médecins que leur mission de conseil auprès du patient. L'évaluation critique de l'IA et le jugement éthique s'établiront comme des compétences essentielles pour tous et, plus encore, pour les médecins. Le défi sera de développer des stratégies pour évaluer les algorithmes qui ne seront pas totalement «compréhensibles» pour l'intelligence humaine.

Pour Werner Bartens, la différence entre la santé et la maladie s'estompe au point qu'une personne en bonne santé est simplement quelqu'un qui n'a pas été examiné avec suffisamment de détail. Diverses méthodes de diagnostic telles que les analyses sanguines et l'imagerie médicale ne sont ni assez sensibles ni assez spécifiques pour mener à des décisions cliniques éclairées. La médecine se concentre sur des solutions techniques et sur des prévisions en apparence exactes, au détriment de compétences telles que la communication et l'empathie, qui ont démontré leur importance dans le processus de guérison.

Le 30 avril 2018, le groupe de travail du CSS a rencontré Torsten Schwede, président du Scientific Expert Board et directeur du Data Coordination Centre du SPHN, et Christian Lovis, chef du service des sciences de l'information médicale à l'Université de Genève et membre du SPHN Scientific Expert Board, afin de discuter de la situation de la conservation des données médicales en Suisse.

Pour Torsten Schwede, l'approche traditionnelle consistant à édifier de grandes bases de données centralisées est inadaptée pour répondre aux défis futurs en raison des questions de confidentialité, des limites techniques et du caractère dynamique de l'acquisition et de l'interprétation de données cliniques dans les hôpitaux modernes. Plutôt que centraliser les données et de rendre obligatoire l'utilisation d'un logiciel spécifique, tous les acteurs devraient accepter et partager des règles et normes communes pour l'annotation et l'interprétation des données.

Pour Christian Lovis, la Suisse pourrait tirer profit de la forte densité de son paysage de données de soins de santé pour explorer de nouvelles pistes de recherche. L'écosystème actuel des données de la recherche biomédicale se caractérise par deux aspects: premièrement, l'utilisation de «données du monde réel» acquises dans des environnements non contrôlés implique le développement de nouvelles approches d'analyse et de validation. Deuxièmement, la coopération et le partage de données nécessitent un cadre commun de sémantique clinique et, au niveau national, l'établissement d'un centre dédié (virtuel ou physique). Il faut noter qu'il existe un retard dans les compétences en matière d'informatique/de phénotype clinique et de sémantique de la santé en Suisse. Il est urgent de développer des capacités dans ce champ scientifique et de l'introduire dans les programmes de formation qui existent déjà.

Les discussions étaient enrichies et renforcées par des rapports d'experts. Sabina Leonelli de l'Université d'Exeter a été invitée par le Conseil à analyser la fonction du *big data* dans le domaine biomédical[17], et Dominic Murphy de l'Université de Sydney a été convié à discuter des différentes positions philosophiques sur la santé et la maladie[18].

Pour Sabina Leonelli, les «données» ne sont jamais dépourvues de contexte scientifique particulier, qu'il s'agisse de la perspective du chercheur à l'origine de leur collecte ou de l'objet de la recherche menée par un utilisateur secondaire. Par conséquent, les cadres théoriques de différents sous-domaines biomédicaux, et notamment leur évolution scientifique continue, doivent être pris en compte lors de la création et de la gestion de bases de données. Étant donné le degré élevé d'interconnexion, le mauvais entretien d'une seule base de données peut affecter la qualité des données à un niveau systémique. Ce risque souligne la nécessité d'un financement durable d'activités chronophages telles que la gestion des données.

Pour Dominic Murphy, il n'existe pas de théorie générale de la santé sur laquelle fonder une définition globale. Dans l'ensemble, la notion de maladie a été étudiée bien plus minutieusement que la notion de santé. Dans les sciences naturelles, la maladie se définit habituellement comme un dysfonctionnement de la nature qui peut être identifié et mesuré de manière objective. D'autres perspectives mettent l'accent sur l'appréciation préalable qu'un état déterminé est nuisible et donc identifié comme une maladie, menant éventuellement à une enquête scientifique. L'usage croissant du *big data* dans les sciences et techniques ne remettra pas en question la conception dominante de la santé en tant qu'objet naturel, mais pourrait accroître le nombre d'états jugés pathologiques.

17   S. Leonelli (2017). La production de connaissances biomédicales à l'ère du Big Data. Analyse réalisée sur mandat du Conseil suisse de la science et de l'innovation. Étude exploratoire 2/2017.

18   Voir annexe du présent rapport.

4

Points de vue sur la santé

L'absence de consensus sur ce que recouvre la notion de santé est une réalité bien connue. La définition formulée en 1948 par l'Organisation mondiale de la Santé (OMS) selon laquelle «la santé est un état de complet bien-être physique, mental et social» est l'une des plus célèbres, mais elle est aussi controversée. La contrepartie de la santé, la maladie, est définie par des cadres de référence tels que la Classification internationale des maladies (CIM)[19] via des classes de maladies distinctes. En Suisse, le concept de santé n'est pas défini par la loi, mais il est utilisé pour délimiter, parmi d'autres concepts, celui de maladie comme «toute atteinte à la santé physique, mentale ou psychique qui n'est pas due à un accident [...] ou provoque une incapacité de travail»[20]. La relation entre santé et maladie peut également varier. Certaines conceptions envisagent une dichotomie entre deux états qui s'excluent mutuellement, tandis que d'autres les envisagent comme les deux extrémités d'un même spectre.

Une majorité de tentatives de définition de la santé s'appuient sur l'une des dimensions suivantes: absence de maladie, bien-être, fonctionnement, homéostasie, flexibilité et/ou capacité à s'adapter[21]. Afin de représenter le champ de réflexion du CSS de manière systématique, le chapitre qui suit explore quatre points de vue sur la santé dans le contexte du *big data*, de la médecine personnalisée et de l'IA en abordant le sujet sous l'angle scientifique, éthique, économique et professionnel. Les deux premières perspectives, la santé comme probabilité et la santé comme valeur, reflètent une vision interne des notions de santé tandis que les deux dernières, la santé comme marché et la santé comme profession, étudient les implications de ces notions pertinentes pour le système suisse de la formation, de la recherche et de l'innovation (FRI) et le système des soins de santé.

4.1

La santé comme probabilité

La santé est de plus en plus exprimée en termes de données et perçue comme le résultat d'opérations computationnelles. Ce point de vue engendre deux types de défis: l'un a trait à la validité des données et des calculs (voir 5.1.1), l'autre à l'interprétation et à la compréhension des données. En raison de l'accumulation croissante des images à haute définition du corps humain, des anomalies structurelles sont relevées même si l'on ignore leur importance pour la santé. Des génomes entiers sont décodés à grande vitesse, mais il n'existe pas de mode d'emploi pour interpréter les données au niveau systémique. Les variantes génétiques, même les rares qui sont bien comprises, ne définissent pas de maladies, mais un certain niveau de risque[22]. Le dépistage peut identifier les personnes à risque avant qu'elles ne développent une maladie. Par exemple, la détection de troubles métaboliques rares chez tous les nouveau-nés empêche quelques-uns d'entre eux de tomber très malades. Toutefois, le dépistage précoce n'améliore pas toujours la survie[23]. De plus, de nouveaux problèmes peuvent être créés au nom de la prévention, en surestimant les risques associés à un trouble ou en sous-estimant ceux causés par une intervention[24]. En fin de compte, alors qu'un risque donné peut être identifié, il n'y a pas d'évidence empirique suffisante pour déterminer le niveau de menace acceptable pour un individu ou une société.

La notion de risque est difficile à saisir pour la majorité des patients, et plusieurs sondages indiquent qu'une partie des médecins n'est pas en mesure de les aider à y voir plus clair. Selon C. Martyn: «Lorsque des médecins proposent un médicament préventif ou un test de dépistage à un grand nombre de personnes asymptomatiques, ils posent un acte totalement différent de celui de traiter un patient venu demander de l'aide parce qu'il est malade. [...] Dans ce nouveau type de médecine, ne pas comprendre le risque revient à ne rien connaître à la circulation sanguine.»[25]

19   L'OMS coordonne à la fois la CIM et l'ICF (Classification internationale du fonctionnement, du handicap et de la santé). Un autre exemple de cadre de référence est le DSM (Manuel diagnostique et statistique des troubles mentaux) publié par l'American Psychiatric Association.

20   LPGA art. 3 al. 1.

21   A. Franke (2012). Modelle von Gesundheit und Krankheit. Dritte überarbeitete Auflage. Verlag Hans Huber, Berne.

22   Voir annexe du présent rapport.

23   H. G. Welch (2018). The heterogeneity of cancer. Breast Cancer Research and Treatment. Vol. 169, Issue 2, pp. 207–208.

24   Voir par exemple: US Preventive Services Task Force (2017). Hormone Therapy for the Primary Prevention of Chronic Conditions in Postmenopausal Women. JAMA. 2017;318(22):2224–2233.

25   C. Martyn (2014). Risky business: doctors' understanding of statistics. BMJ 2014;349:g5619.

Prédire l'autisme

L'autisme est un trouble du développement touchant 17 enfants américains sur 1000 selon des estimations récentes. Ses causes ne sont pas bien comprises. Les indicateurs de prédisposition génétique sont variés, et le dépistage chez le tout jeune enfant est très délicat. À l'âge de 2 ans, les enfants autistes présentent des schémas comportementaux de retrait social bien reconnaissables. En 2017, une équipe américaine a annoncé pouvoir diagnostiquer l'autisme chez des bébés de 6 mois avec 81% de précision et 88% de sensibilité à l'aide de scanners cérébraux et de l'apprentissage profond (*deep learning*)[26]. Peu de temps après, des performances encore meilleures ont été enregistrées en analysant des tracés d'électroencéphalogramme chez des bébés de 3 mois, avec une précision et une sensibilité de 95%[27].

Une précision de 95% apparaît nettement supérieure aux méthodes actuellement établies. Pourtant, la proportion de faux positifs plaide pour réserver le test aux nourrissons déjà jugés à risque: baser la prédiction sur le seul résultat positif du test signifie que la probabilité de l'enfant à être autiste à l'âge de 2 ans serait seulement de 25%[28] (et pas plus de 7% dans le test d'imagerie cérébrale le moins précis). Il est probable qu'une intervention précoce atténue les symptômes ultérieurs, apportant ainsi un véritable bénéfice aux enfants touchés, tandis qu'un faux positif causera probablement du tort au développement de l'enfant et à sa relation avec sa famille. Dans le meilleur des cas, si un traitement précoce aide effectivement à prévenir les difficultés sociales, il peut même devenir impossible de faire la différence ultérieurement entre les «vrais» et «faux» positifs, notamment parce que l'autisme est considéré comme un spectre et non une condition distincte.

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26   H. C. Hazlett et al. (2017), Early brain development in infants at high risk for autism spectrum disorder. Nature 2017; 542(7641):348-351.

27   W. J. Bosl et al. (2018). Early brain development in infants at high risk for autism spectrum disorder. Sci Rep. 2018 1;8(1):6828.

28   Sur 1000 enfants, 934 seront négatifs avec un résultat négatif correct, 16 seront autistes avec un résultat positif correct, 1 sera autiste mais faux négatif et 49 seront de faux positifs.

4.2

La santé comme valeur

Si l'on se base sur le montant que les citoyens suisses sont disposés à investir, à la fois en fonds publics et privés[29], il semblerait que la santé – et la sécurité en général – est considérée comme le bien le plus précieux. Pourtant, l'attitude de nombreuses personnes indique qu'elles considèrent leur santé comme un bien relatif, par exemple en donnant la priorité à leur carrière ou en se donnant la liberté de ne pas toujours faire le «bon choix» pour leur santé. Ces valeurs et leurs ramifications éthiques façonnent le système suisse des assurances sociales. La santé, après tout, est aussi un bien public au sens où même les personnes les plus riches et jouissant d'une excellente santé gagnent en sécurité si d'autres membres de la société souffrent de moins de maladies infectieuses, de troubles mentaux ou d'addictions[30].

Aujourd'hui, la tendance à considérer la santé comme étant essentiellement le fruit d'un comportement individuel gagne du terrain. L'approche de médecine personnalisée et le mouvement du *quantified self* se concentrent sur l'individu par opposition à la société comme la source de données et d'affinement des prédictions. L'espoir que la technologie puisse offrir de meilleures armes à l'individu remet en question l'adhésion générale de la société au principe de solidarité[31]. Dans le contexte de la disponibilité et de l'ubiquité générales des données, une manifestation importante du conflit éthique entre solidarité et autonomie sera de savoir si l'actuel «droit de ne pas savoir» (voir encadré page suivante) pourra être préservé à l'avenir.

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29   En 2016, les dépenses globales de soins de santé atteignaient CHF 80,7 milliards ou 12% du produit intérieur brut de la Suisse. En 1996, le même poste représentait CHF 37,5 milliards ou 9% du PIB national.

30   En outre, un argument économique pourrait être avancé en termes d'augmentation des prestations sociales et de pertes de paiement d'impôts.

31   F. Gutzwiller et A. Müller (2017). Gerechtigkeit im digitalen Zeitalter. Personalisierte Daten – eine Frage der Solidarität. NZZ. 18.10.2017.

Le droit de ne pas savoir

Une grande proportion de patients susceptibles de développer la maladie de Huntington préfère ne pas se soumettre au dépistage génétique[32]. Clairement, la certitude de développer la maladie est susceptible de dégrader la qualité de vie psychologique des années avant la survenue de celle-ci. De plus, même pour les porteurs d'une mutation autosomique dominante telle que Huntington, la prédiction clinique reste imprécise puisque la gravité, l'âge d'apparition de la maladie et la rapidité de progression varient. Dans tous les cas, la décision de se soumettre ou non à un test génétique n'est jamais prise sur la base de facteurs strictement personnels: les enfants, les frères et sœurs et même des parents éloignés peuvent être concernés par le résultat d'un test qu'ils n'ont pas demandé.

La conception de solidarité due à la famille varie d'un pays à l'autre. Selon la législation française, une personne qui apprend un risque génétique est tenue d'informer sa famille, bien qu'elle puisse demander à son médecin traitant de le faire à sa place (loi relative à la bioéthique du 7 juillet 2011, art. 2). En Suisse, il est conseillé à cette même personne d'informer sa famille sans que son refus d'agir de la sorte n'entraîne de conséquence pour elle. Dans ce cas toutefois, le médecin traitant doit décider si l'intérêt de la famille du patient prévaut sur son devoir de confidentialité vis-à-vis de son patient.

Dans un souci d'autonomie personnelle, l'article 8 de la loi fédérale sur l'analyse génétique humaine (dans sa version de juin 2018) stipule que «toute personne peut refuser que tout ou partie des informations relatives à son patrimoine génétique lui soit communiquée». Le «droit de ne pas savoir» est ainsi l'expression d'un souhait actif et explicite de la personne concernée. Les circonstances de son application pourraient devenir plus fréquentes, y compris dans les situations dépassant le cadre génétique, étant donné le nombre croissant d'algorithmes prédictifs. Naturellement, sa faisabilité reste limitée, car un patient doit savoir qu'il est à risque avant de pouvoir décider de recevoir ou non l'information.

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32   P. J. Morrison (2010). Accurate prevalence and uptake of testing
      for Huntington's disease. Lancet Neurol. 2010; 9(12):1147

La santé comme marché

L'amélioration de leur santé est le seul objectif pour lequel les personnes n'envisagent pas de limiter leurs efforts. C'est pourquoi les soins de santé constituent le secteur de croissance par excellence, offrant des emplois et des opportunités d'exportation, et confrontant les finances publiques à des augmentations de coûts de plus en plus problématiques[33]. Le marché des soins de santé est exposé à des défaillances en raison de ses caractéristiques uniques telles que de fausses incitations et le niveau d'information très inégal entre prestataires et consommateurs[34]. Les médecins se soumettent eux-mêmes à moins de traitements médicaux que ce qu'ils recommandent aux patients[35]: ils sont sans doute les mieux placés pour réaliser qu'un traitement plus poussé ne signifie pas automatiquement de meilleurs résultats, alors que le système actuel récompense le traitement, et non son aboutissement. Dans les hôpitaux suisses, les bonus annuels pour la réalisation d'un nombre minimal d'interventions sont fréquents. Par conséquent, de nombreux patients, surtout ceux ayant souscrit une assurance privée, sont incités à subir des procédures inutiles, courant par là même le risque d'effets indésirables et secondaires[36]. Les médecins en cabinets privés sont mieux récompensés pour les interventions techniques que pour leurs activités intellectuelles et de communication.

Le monde politique exerce une forte pression pour réduire les coûts du système d'assurance invalidité en promouvant la réinsertion et en compliquant progressivement l'accès aux prestations d'invalidité pour les nouveaux demandeurs. La pression est un peu plus faible dans le système d'assurance santé général parce que les électeurs rejettent généralement les propositions qui limitent leur éventail d'options de soins. Plusieurs conflits d'intérêts rendent les changements systémiques très difficiles, que ce soit au niveau des citoyens

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33   Voir note 29, p. 17.

34   OFSP (2017). Mesures visant à freiner la hausse des coûts dans l'AOS.
      Rapport du groupe d'experts du 24.08.2017.

35   K. Murray (2011). How Doctors Die. Zócalo. Disponible sous:
      http://www.zocalopublicsquare.org/2011/11/30/how-doctors-die/
      ideas/nexus/; V. S. Periyakoil et al. (2014). Do Unto Others: Doctors'
      Personal End-of-Life Resuscitation Preferences and Their Attitudes
      toward Advance Directives, PLOS.

36   O. Peters et al. (2016). Excédent de prestations stationnaires pour les
      personnes avec une assurance complémentaire. OFSP, 10.06.2016;
      F. Gottschalk et al. (2017). Health Services as Credence Goods: A Field
      Experiment. EPF Zurich, septembre 2017.

(voir par exemple: l'initiative sur la caisse unique, le référendum sur les soins intégrés, les décisions locales relatives à la fermeture d'hôpitaux), des membres du Parlement (votes sur la prévention, les réglementations sur le tabac) ou des prestataires de soins de santé (négociations tarifaires TARMED)[37]. Si la tendance actuelle se poursuit, le coût des soins de santé pourrait augmenter à un point tel que la santé de la population suisse se trouverait affectée par la limitation des dépenses discrétionnaires pour d'autres tâches publiques, en particulier au niveau cantonal.

4.4
La santé comme profession

Les médecins sont les acteurs clés du système de santé. C'est à eux que revient la responsabilité de mettre un nom sur un trouble et d'évaluer si une personne est malade et doit bénéficier d'un traitement ou d'une période d'incapacité de travail. Par exemple, un expert médical mandaté par l'assurance invalidité détermine si une personne – qui n'est pas son patient – n'est plus en mesure de travailler, ce qui s'avère une tâche complexe dans le cas de douleurs chroniques ou de troubles mentaux. Dans un cabinet médical, cependant, le problème le plus fréquent n'est pas le manque d'«évidences» mesurables, mais l'absence de formule toute prête pour en extraire du savoir. Au vu du rythme auquel les séquences génétiques et images en 3D sont générées, il est souvent difficile de dire quel type de médecin, du généraliste aux différents spécialistes, doit interpréter quelle information[38]. Même pour les maladies et les tests en laboratoire courants, l'usage sémantique des données et des descriptions de symptômes n'est pas uniformisé entre les hôpitaux[39]. De nos jours, les patients se rendent souvent au cabinet de leur médecin avec leur propre hypothèse de diagnostic. Chaque patient et chaque patiente possède son propre objectif en termes de santé, qui n'est pas systématiquement en phase avec celui du médecin.

Selon une présupposition généralement admise, les médecins de demain auront besoin d'un éventail de compétences encore plus vaste qu'actuellement: davantage de physique et de biochimie, de meilleures compétences sociales et en communication, une maîtrise approfondie des probabilités, des notions de médecine alternative et, si possible, des connaissances en informatique, codage et ingénierie médicale. Le résultat pourrait être une surcharge de la formation médicale, ce qui pourrait amener, malgré les efforts déployés, à se concentrer sur l'accumulation de faits et l'enseignement en fonction des tests.

D'autres professionnels aux rôles essentiels pour le système de soins de santé, comme les infirmiers, abandonnent massivement le métier, démontrant par là qu'attirer et former plus de candidats n'est pas suffisant pour garantir le bon nombre de prestataires de soins[40]. Il y a des raisons de croire qu'avec de meilleures conditions de travail, bon nombre de ces prestataires seraient restés dans la profession[41]. Les Suisses seront invités à voter sur une initiative populaire proposant, entre autres choses, la redistribution de certaines compétences entre les médecins et les infirmiers[42].

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37   Si elles aboutissent, les discussions politiques permanentes visant à harmoniser les modes de paiement pourraient décourager les hôpitaux à dispenser des traitements hospitaliers plutôt que des soins ambulatoires. Cette mesure ne résoudrait toutefois pas le problème de préférence de la quantité de traitements à la qualité des soins.

38   R. R. Stavert & J. P. Lott (2013). The Bystander Effect in Medical Care. NEJM. 2017. 2013(368):8-9.

39   A. K. Manrai, C. J. Patel, J. P. A. Ioannidis (2018). In the era of precision medicine and big data, who is normal? JAMA. 15 May 2018, Volume 319, Number 1.

40   En 2013, 32% des infirmiers de 35 ans et moins avaient abandonné le métier qu'ils avaient appris. Cette proportion grimpe à 56% pour les infirmiers de 50 ans et plus. (M. Lobsiger et al. ‹2016›. Les professionnels de la santé: sorties de leur profession. Obsan Bulletin 7/2016, Neuchâtel.)

41   R. Schwendimann et al. (2014). Das Pflegefachpersonal in Schweizer Spitälern im europäischen Vergleich. Obsan Bulletin 3/2014, Neuchâtel; C. Merçay et al. (2016). Personnel de santé en Suisse. Etat des lieux et projections à l'horizon 2030. Obsan Rapport 71, Neuchâtel.

42   https://www.bk.admin.ch/ch/f/pore/vi/vis472t.html.

5
Position du CSS

5.1
Synthèse

Sur la base de contributions externes et de perspectives internes présentées dans les chapitres précédents, le Conseil propose d'agir dans les domaines de la recherche et de l'innovation, de la formation et de la promotion de la santé. Le CSS considère sa position comme complémentaire à d'autres voix contribuant à des thèmes importants tels que la limitation des coûts[43], la gouvernance des données[44], l'éthique[45] ou la qualité de vie[46], pour n'en citer que quelques-uns.

5.1.1
Recherche et innovation

Les approches analytiques exploitant de grands ensembles de données ouvrent une série de nouvelles possibilités pour la science et la médecine. Leur potentiel est évident dans le développement de nouveaux diagnostics, et il est en train de s'établir en ce qui concerne la prise de décision assistée entre différentes options thérapeutiques. Ces approches peuvent aussi mener à une meilleure compréhension des mécanismes pathologiques grâce à l'identification de corrélations insoupçonnées. Mais pour établir des relations de cause à effet, la confirmation par l'expérimentation reste indispensable. Étant donné la propension des experts et non-experts à considérer les interventions onéreuses comme supérieures aux alternatives moins chères[47], les innovations dans les soins de santé doivent démontrer leur supériorité sur la base de preuves cliniques.

Les principaux problèmes d'importance scientifique se rapportent à (i) la qualité du traitement des données pendant leur collecte, leur annotation et leur conservation, (ii) la publication de résultats de recherche dits négatifs et (iii) la reproductibilité

des expériences publiées. Si ces défis ne sont nullement spécifiques aux approches de recherche faisant appel au *big data*, il est peu probable qu'on puisse les régler sans les aborder explicitement. Regrouper plusieurs ensembles de données non représentatifs, loin de garantir une compensation des biais respectifs, augmente plutôt les problèmes de reproductibilité en raison des limites techniques et du manque d'accès aux données ou aux algorithmes[48]. Ces problèmes de validité scientifique peuvent expliquer pourquoi nombre de chercheurs n'utilisent que les données de collègues qu'ils connaissent personnellement. Le partage de données à plus grande échelle ne peut donc réussir que grâce à la définition et l'utilisation systématique de terminologies sémantiques et de normes de traitement. Étant donné que ces normes évoluent continuellement sur le plan scientifique, un centre ou institut national de coordination est nécessaire pour évaluer et veiller à l'application de meilleurs standards.

5.1.2
Formation

Le *big data* est un prérequis à l'apprentissage automatique et à l'IA, qui transformeront la pratique de la médecine. Les compétences que devront posséder les professionnels des soins de santé de demain vont continuer d'évoluer. La pensée synthétique et la déduction logique gagnent en importance, tandis que les connaissances encyclopédiques ou l'accumulation de faits sont progressivement déléguées aux systèmes d'assistance. De fait, les machines prennent en charge un nombre croissant d'essais et actes techniques actuellement réalisés par des médecins. Les médecins de demain devront aussi développer des compétences approfondies en évaluation critique de l'IA et maîtriser tant le langage des algorithmes que la communication verbale et non verbale avec le patient. Et ils devront par-dessus tout pouvoir saisir la perspective unique du patient afin de l'aider à se retrouver dans la complexité croissante des options de soins. Ces perspectives entraîneront une adaptation de la formation médicale.

5.1.3
Promotion de la santé

L'analyse philosophique[49] et le débat des experts[50] parviennent tous deux à la conclusion que la numérisation et le *big data* ne peuvent libérer la notion de santé de sa nature à la fois subjective et objective. Il n'existe pas de notion de santé socialement

43   OFSP (2017). Mesures visant à freiner la hausse des coûts dans l'AOS. Rapport du groupe d'experts du 24.08.2017.

44   B. Prainsack (2015). Personalized medicine. Empowered patients in the 21st century? NYU Press. Series: Biopolitics, décembre 2017.

45   European Commission Science Advice and Ethics Groups at ESOF (2016). Ethics of New Health Technologies and Citizen Participation. Opinion n°29 - 13/10/2015; European Group on Ethics in Science and New Technologies (2018). Statement on the Ethics of Artificial Intelligence, Robotics and "Autonomous Systems"; Ethical Framework for Responsible Data Processing within SPHN (Version 1, 12.06.2017).

46   ASSH (2016). Définir, mesurer et promouvoir la qualité de vie. Lebensqualitätsforschung in der Schweiz.

47   R. L. Waber et al. (2008). Commercial features of placebo and therapeutic efficacy. JAMA. 299, 1016–1017. / A. J. Espay (2015). Placebo effect of medication cost in Parkinson disease: a randomized double-blind study. Neurology. 2015 Feb 24;84(8):794-802.

48   J. T. Wilbank et E. J. Topol (2016). Stop the privatization of health data. Nature. Vol. 535, July 2016.

49   Voir annexe du présent rapport.

50   ASSM & CSS (2019). Patient, médecin, big data. Qui a le pouvoir de définition? Swiss Academies Communications. Vol. 14, No 3, 2019.

neutre ou exempte de toute valeur. C'est pourquoi la science et la médecine doivent prendre en compte le fait que les préoccupations éthiques et sociales influencent le choix des données et des métadonnées utilisées par les professionnels des soins. Globalement, le *big data* présente deux scénarios possibles pour le système de santé. Il peut élargir encore davantage la vision courante de ce qui relève du pathologique, transformant de plus en plus de personnes en individus à risque et pratiquement chaque trouble en maladie rare[51]. Mais il pourrait aussi favoriser la représentation d'une coexistence possible entre plusieurs modes d'être en bonne santé. Au lieu de se focaliser sur la diversité des risques pathologiques, la science pourrait étudier les variations entre individus en bonne santé, pour avancer l'idée que ne pas correspondre à une norme physiologique ou psychologique n'équivaut pas automatiquement à être malade.

5.2
Recommandations du CSS et commentaire

5.2.1
Promouvoir une compréhension globale de la santé

1: Le CSS invite le Conseil fédéral à considérer la formation de base, les programmes sociaux et les questions environnementales comme des investissements prioritaires en tant que mesures de promotion de la santé à part entière.

La santé n'étant pas une notion purement objective, certaines approximations sont nécessaires pour en parler de manière quantitative; la longévité est un de ces indicateurs. Considérer l'espérance de vie moyenne comme un «proxy» du niveau de santé n'implique pas que prolonger la vie et améliorer la santé soient toujours synonymes, mais signifie que des informations statistiques peuvent renseigner sur les conditions les plus favorables à la santé[52]. Des études sur la longévité ont montré que les soins de santé ne sont pas, et de loin, le seul paramètre contribuant à la santé.

Pourquoi vivons-nous plus longtemps?

Durant la période suivant la Seconde Guerre mondiale, le recours à la science pour résoudre des problèmes de santé majeurs, comme la tuberculose, la polio ou la rougeole, a suscité un optimisme justifié. Thomas McKeown (1912–1988), un médecin britannique, a remis en question l'idée selon laquelle les soins médicaux sont le principal vecteur de l'espérance de vie, soulignant le rôle de la croissance économique, de l'augmentation du niveau de vie et de l'amélioration de la nutrition. Il a observé qu'une grande partie de la baisse de mortalité due aux principales maladies infectieuses avait eu lieu avant la découverte et l'administration des antibiotiques. Les affirmations de McKeown ont été contestées ultérieurement. Aujourd'hui, l'OMS explique le fait qu'un enfant né en 2018 soit susceptible d'atteindre un âge avancé dans presque tous les pays par les facteurs suivants: la nutrition, l'hygiène, le niveau d'éducation de la mère et l'accès aux techniques médicales, surtout les vaccins et les antibiotiques. Le progrès scientifique est à la base de pratiquement tous ces aspects, mais les soins de santé en tant que tels ne constituent qu'une partie de l'équation.

Le statut socioéconomique reste lié à la longévité, même dans les pays à revenus élevés. En Suisse, la différence d'espérance de vie entre habitants de quartiers riches ou pauvres d'une même ville est de 4,5 ans pour les hommes et 2,5 ans pour les femmes[53]. Au Royaume-Uni, l'écart correspondant est de 9 ans pour les hommes et 7 ans pour les femmes[54].

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53 OFSP (2018). Égalité des chances et santé. Chiffres et données pour la Suisse.

54 Public Health England (2017). Reducing health inequalities: system, scale and sustainability.

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51 Voir annexe.

52 C'est particulièrement le cas lors de l'examen des tendances du recul de l'espérance de vie. Voir par exemple: S. H. Woolf et L. Y. Aron (2013). The US health disadvantage relative to other high-income countries: findings from a National Research Council/Institute of Medicine report. JAMA. 2013; 309:771; S. H. Woolf et al. (2018). Changes in midlife death rates across racial and ethnic groups in the United States: systematic analysis of vital statistics. BMJ. 2018; 362:k3096.

Différents modèles expliquent la diversité des facteurs déterminant la santé, et des recherches plus approfondies sont nécessaires pour comprendre leurs contributions respectives[55]. Toutefois, selon un consensus établi au cours de la dernière décennie, les déterminants structurels – soit les facteurs qui ne peuvent être facilement modifiés par les individus, comme l'habitat et les conditions de travail – sont plus importants que des facteurs comportementaux tels que le régime ou l'exercice physique[56]. Sans nier l'espoir que les patients et les citoyens tirent profit des nouvelles connaissances et technologies pour améliorer leur propre santé, il semble clair qu'une modification des comportements ne suffit pas et qu'il faut des politiques englobant protection sociale, politique fiscale, réglementation sur le lieu de travail, environnement, sécurité des transports, etc. Un enseignement de qualité est largement considéré comme l'un des principaux facteurs de santé[57]. Tous les efforts doivent être déployés pour éviter que les autorités publiques ne soient contraintes de diminuer leurs investissements dans l'enseignement de base pour des raisons budgétaires à courte vue.

De plus, des programmes préventifs efficaces devraient être développés, non pas dans l'espoir – pas toujours justifié – qu'ils réduisent les coûts, mais en raison de leur haut potentiel avéré d'amélioration de la santé[58]. Les programmes préventifs couvrent une grande variété de mesures. Dans l'exemple de la prévention de l'obésité, R. Z. Goetzel propose ceci: «Les normes sociales peuvent être significativement modifiées en coordonnant les efforts impliquant de multiples acteurs (par exemple, urbanistes, cultivateurs et distributeurs, éducateurs, représentants du gouvernement, responsables sociaux et professionnels des soins de santé) sans compter exclusivement sur le personnel médical onéreux pour améliorer la santé.»[59]

2:    Le CSS recommande à l'OFSP et au SEFRI de soutenir durablement la recherche en matière de services de santé.

La notion de *big data* renvoie à des ordres de grandeur qui ont différé selon les périodes historiques[60]. Dans ce sens, la science a travaillé avec de grands ensembles de données complexes depuis des décennies, y compris dans des domaines tels que l'épidémiologie et la santé publique. À présent, le *big data* dans le contexte biomédical implique généralement une combinaison de génomique, de protéomique ou de métabolomique. Des mécanismes doivent être renforcés et d'autres créés pour permettre l'intégration fructueuse de telles approches moléculaires avec d'autres formes de recherche. Différents types d'informations, comme les données des assureurs, les cohortes de recherche ou les études environnementales, ainsi que les résultats d'études épidémiologiques, doivent être intégrés dans les algorithmes et processus d'évaluation de la santé. Les méthodes qui permettent le suivi et l'observation de sujets volontaires dans le «monde réel», une approche facilitée par les nouveaux dispositifs technologiques, seront d'une utilité particulière.

La recherche en matière de services de santé – c'est-à-dire le domaine scientifique visant à identifier des solutions efficaces pour organiser, financer et fournir des soins de qualité – a vu le jour il y a 40 ans aux États-Unis. Cette forme de connaissance scientifique est cruciale pour déterminer quand et où appliquer de manière prudente et différenciée le concept du «moins, c'est plus»[61]. En Suisse, les premières étapes ont été mises en place avec le financement initial par l'ASSM et la Fondation Gottfried et Julia Bangerter-Rhyner (2012–2016), et ensuite via le Programme national de recherche (PNR) 74 «Système de santé» (2017–2022). Un soutien restera nécessaire à l'issue du PNR, car ce domaine de recherche doit continuer à croître. À ce stade, la plupart des études dans la recherche en matière de services de santé décrivent des variations en qualité et en efficacité. Fournir des théories de causalité et des modèles déductifs renforcerait encore davantage la valeur de ces recherches.

3:    Le CSS recommande au SEFRI et à l'OFSP d'encourager un débat continu pour déterminer les attentes de la société en matière de soins de santé et de qualité de vie.

La définition de la santé intègre de nombreux paramètres avec une grande complexité. L'éventail toujours plus large d'options de diagnostic disponibles pourrait aboutir à délimiter une norme très étroite, renforçant la tendance à la surmédicalisation déjà observée dans les sociétés à haut revenu. S'appuyant sur des initiatives privées existantes[62], les autorités publiques devraient soutenir le développement de nouvelles plateformes d'échange interdisciplinaires et impliquer le public et les chercheurs pour déterminer si les soins de santé doivent être appelés à prolonger la vie à tout prix.

55   N. Cantoreggi (2010). Pondération des déterminants de la santé en Suisse: Etude réalisée dans le cadre de l'élaboration d'un modèle de déterminants de la santé pour la Suisse. Université de Genève.

56   M. Richter et al. (2011). Determinanten der Gesundheit und ihre relative Bedeutung für die Erklärung gesundheitlicher Ungleichheiten: Ein systematisches Review. Université de Berne.

57   OFSP (2018). Égalité des chances et santé. Chiffres et données pour la Suisse.

58   A. E. Carroll (2018). Preventive Care Saves Money? Sorry, It's Too Good to Be True. The New York Times. 29.01.2018.

59   R. Z. Goetzel (2009). Do Prevention Or Treatment Services Save Money? The Wrong Debate. Health Affairs. Vol. 28, No. 1: The Crisis In Chronic Disease

60   S. Leonelli (2017). La production de connaissances biomédicales à l'ère du Big Data. Analyse réalisée sur mandat du Conseil suisse de la science et de l'innovation CSSI. Étude exploratoire 2/2017.

61   A. Gawande (2015). Overkill. The New Yorker. 11.05.2015.

62   TA-SWISS, Science et Cité, Leenards Foundation et miDATA pour n'en citer que quelques-unes.

Au sein du corps médical, des mouvements tels que l'association Choosing Wisely ont mis sur la table des réflexions sur les coûts et bénéfices des interventions médicales. Ces échanges débouchent souvent sur des débats difficiles mais importants sur les limites et la valeur de la vie humaine[63]. Inclure les perspectives des sciences naturelles, de l'ingénierie, des sciences sociales et des sciences humaines est une étape nécessaire mais insuffisante pour parvenir à établir un consensus sur la santé. Afin de concevoir une notion de santé acceptable pour la société suisse et de déterminer jusqu'à quel point l'analyse des données quantitatives peut contribuer à la cerner, il faudrait impliquer des professionnels des soins de santé, des patients et des particuliers dans un plus large débat sur le but des soins de santé[64].

5.2.2
Développer des standards de données favorables à la santé

4: *Le CSS recommande à l'OFSP de réduire le biais de publication dans la littérature clinique en assurant l'accès aux résultats des essais cliniques.*

Le *data mining* à grande échelle repose sur la littérature scientifique et sur des bases de données en *open source* afin d'obtenir la base d'évaluation la plus large possible. Grâce aux réglementations imposant l'enregistrement de tous les essais cliniques dès leurs débuts (depuis 2014 en Suisse avec la loi relative à la recherche sur l'être humain), il est possible d'estimer le pourcentage d'études réalisées et publiées, qui varie selon les pays et les promoteurs de recherche, mais se limite en moyenne à seulement une étude publiée sur deux[65]. L'absence de rapport sur les résultats d'un essai peut être le fait d'un abandon pour des problèmes de faisabilité. Mais le chercheur peut aussi choisir de ne pas publier parce que les résultats de son étude n'étayent pas l'hypothèse initiale. Les personnes qui se portent volontaires doivent pouvoir espérer que leur participation à un essai clinique contribue à faire progresser la science indépendamment des intérêts propres des promoteurs de la recherche. Même les études interrompues pour cause de recrutement insuffisant ou pour d'autres raisons techniques méritent d'être

rapportées, et ce, pas forcément sous la forme d'une publication classique, afin d'éviter que des projets cliniques futurs ne répètent les erreurs du passé. La Confédération devrait encourager la publication resp. la mise à disposition des résultats de tous les essais cliniques en faisant prendre conscience de ce problème au niveau international, en garantissant une description précise des métadonnées des dits essais et en finançant les bases de données et/ou les nouveaux formats de publication nécessaires à la communication de résultats négatifs.

5: *Le CSS recommande à l'OFSP d'augmenter les compétences et de définir les critères de validation pour les logiciels médicaux.*

Si le biais de publication dans la littérature scientifique est un problème reconnu, la question de l'efficacité clinique et des répercussions sociales de l'IA dans le domaine des soins de santé est relativement récente. Selon la loi sur les produits thérapeutiques, les applications, instruments autonomes en ligne et les logiciels intégrés aux robots chirurgicaux ou aux appareils d'imagerie sont des dispositifs médicaux devant être agréés par des organes d'accréditation via une procédure simplifiée. Mais les algorithmes actuels sont souvent trop complexes pour être compris par les régulateurs et les utilisateurs. Il est difficile de savoir si les données générées par les nouveaux dispositifs médicaux sont protégées par la loi, et il est de plus en plus compliqué de définir les spécifications d'un système ou d'évaluer la qualité des données utilisées pour l'entraînement des algorithmes d'apprentissage automatique. Les mises à jour, rarement soumises à vérification, peuvent représenter un changement insignifiant ou bien entraîner de profondes répercussions. Le système de réglementation actuel suppose que les hôpitaux et les médecins sont suffisamment informés sur les algorithmes utilisés par leurs instruments: ce point mérite d'être vérifié. Il convient de renforcer l'expertise et de mettre à jour les critères pour affiner l'évaluation des logiciels médicaux – à la fois au niveau national et international – sur le plan de l'efficacité, de la performance technique et eu égard à l'impact social de l'application[66]. Le nouveau centre suggéré par le CSS (voir recommandation 8) pourrait jouer un rôle majeur dans ce domaine.

63   D. Friedli (2018). Ein Leben ist dem Bund plötzlich fast doppelt so viel wert. NZZ am Sonntag. 07.07.2018.

64   Sur les buts de la médecine et du système de santé, voir la Feuille de route de l'ASSM (2019). Développement durable du système de santé; Sur la participation du public à la science, voir: B. Strasser & M. Haklay (2018). Citizen Science: Expertise, Democracy and Public Participation. SSC policy analysis 1/2018.

65   B. Goldacre (2018). Compliance with requirement to report results on the EU Clinical Trials Register: cohort study and web resource. BMJ 2018, 362:k3218; Amstutz, A. et al. (2018). Funding characteristics of randomised clinical trials supported by the Swiss National Science Foundation: a retrospective cohort study. Swiss Med Wkly. 2018; 148.w14587.

66   Voir l'exemple d'IBM Watson pour une discussion du biais culturel dans les logiciels médicaux: Ross, C. et I. Swetlitz (2017). IBM pitched its Watson supercomputer as a revolution in cancer care. It's nowhere close. STAT. 2018.

6: Le CSS recommande au SEFRI et aux autorités cantonales de soutenir les services essentiels tels que l'annotation de données et la conservation de bases de données.

Garantir un financement durable pour la tâche essentielle d'annotation et de conservation des données est compliqué. Même les activités de l'Institut suisse de bioinformatique relatives à ses ressources les plus utilisées (p. ex. UniProt) dépendent en large part de sources de financement américaines. Le FNS ne considère pas l'annotation des données, sauf cas exceptionnel, comme faisant partie des coûts directs de projet. De nouveaux modèles de financement distribué via des agences de financement de la recherche pourraient être à étudier, notamment pour les sciences fondamentales du vivant[67]. Mais pour ce qui est des données cliniques, il semble logique que les autorités publiques locales assument la responsabilité du financement durable nécessaire à la mise en œuvre de standards d'annotation. Une solution consisterait à encourager les hôpitaux et/ou les hautes écoles suisses à créer et gérer ce service. Alternativement, ce service pourrait être intégré dans un centre national d'informatique médicale (voir recommandation 8). La conservation des bases de données sur un plan local selon les recommandations d'un organe central serait la solution idéale.

7: Le CSS recommande à l'OFSP et au SEFRI de garantir la prise en compte des implications sociales liées au choix des données et à leurs spécifications.

T. Wilbanks et E. Topol déclaraient en 2016: «Si la prise de décision fondée sur des algorithmes commence à infiltrer les données de santé à notre insu, la capacité des calculs secrets à accentuer les biais préexistants dans notre société pourrait augmenter significativement. [...] On peut facilement imaginer un avenir où les entreprises pourront vendre les profils de maladies des patients à leur insu. Ou une société où les décisions de santé sont obscures et invérifiables, et où les progrès dans la compréhension sont utilisés pour la commercialisation agressive de services de soins – que ces services soient effectivement bénéfiques à leur santé ou non.» Les algorithmes prédictifs devraient être utilisés et distribués avec la plus grande prudence et évalués par des experts indépendants qui maîtrisent les questions éthiques et les techniques de l'information.

Seul un petit nombre d'algorithmes liés à la santé sont utilisés dans le système de soins de santé suisse à l'heure actuelle. On peut toutefois imaginer les applications suivantes:

— Des systèmes de prise de décision pour prioriser les patients en attente d'un don d'organe. La médecine de transplantation utilise déjà un «algorithme», soit un ensemble de règles défini au niveau national. Recourir à l'IA pour améliorer cet algorithme serait l'étape suivante.

— Juger si une personne mène une vie saine ou non: les compagnies d'assurances se servent déjà d'algorithmes pour décider si une personne peut souscrire une assurance privée.

— Des modèles prédictifs pour établir les besoins des résidents par rapport aux services sociaux locaux: ce genre d'initiative de santé publique pourrait être compliqué par le manque de représentativité puisque les personnes sans ressources tendent à générer moins de données.

— La détection et la prévision de dynamiques d'infections telles que le sida, Ebola ou des souches de grippe spécifiques: l'épidémiologie pourrait affiner ses modèles à l'aide de l'IA. Mais des prédictions positives erronées peuvent générer de sérieuses répercussions économiques dans les villes et les pays concernés.

— Des scientifiques de l'Université de Stanford ont récemment développé un logiciel pour estimer la durée de vie probable d'une personne et le moment idéal pour engager une discussion sur les soins palliatifs[68]. Toutes les précautions doivent être prises pour s'assurer que de tels algorithmes ne contribuent pas à creuser les inégalités.

En l'absence de prise en considération de l'impact social des algorithmes, les minorités en termes d'âge, de maladie, d'ethnie et de caractéristiques sociologiques courent le risque d'une prise en charge insuffisante et/ou d'une surmédicalisation. De plus, les experts qui déterminent les limites entre le «normal» et le «pathologique», la nature des données à collecter, la manière de les annoter et la façon de partager les résultats pour définir l'ontologie clinique exercent une responsabilité majeure dans la définition de la santé. Leur indépendance vis-à-vis de tous les autres acteurs est capitale.

67   C. Gabella et al. (2017). Funding knowledgebases: Towards a sustainable funding model for the UniProt use case. F1000Research 2018, 6 (ELIXIR):2051.

68   A. T. Hsu (2018). Algorithm for predicting death among older adults in the home care setting: study protocol for the Risk Evaluation for Support: Predictions for Elder-life in the Community Tool (RESPECT). BMJ Open. 2016; 6(12): e013666.

5.2.3
Préciser les objectifs de l'initiative SPHN

8: Le CSS recommande au SEFRI, à l'OFSP et aux autorités cantonales de créer un centre national pour l'informatique médicale et l'analyse des données médicales, qui définira des normes et orientera l'ensemble des institutions médicales suisses dans le contexte de l'initiative SPHN.

La recherche de nouvelles découvertes scientifiques basées sur le *big data* a été décisive dans la mise en place de programmes de recherche de grande envergure couvrant un large éventail d'intérêts scientifiques, du Projet du génome humain (1990–2003) au Human Brain Project (2013–2022), en passant par des initiatives nationales telles que SystemsX.ch (2008–2016) et le Swiss Personalized Health Network (démarré en 2017). Les grands projets de recherche peuvent être considérés comme un point de cristallisation pour la compréhension de concepts biomédicaux majeurs et la réflexion sur leur éventuel impact médical. Pour sa part, le SPHN jette des ponts entre les institutions, les disciplines et les chercheurs pour garantir l'interopérabilité des données de santé.

L'expérience montre qu'une telle mission nécessite une structure de coordination à long terme. C'est pourquoi la prochaine phase du SPHN devrait poursuivre les efforts actuels de constitution et de soutien des compétences au niveau national. Durant la deuxième phase du SPHN, un centre national d'informatique médicale devrait être créé avec un financement durable correspondant à ses fonctions essentielles. Ce centre devrait se concentrer sur la définition de normes et l'évaluation des algorithmes à appliquer dans les établissements médicaux suisses.

À long terme, l'organisation de ce centre suisse d'informatique médicale serait distincte de celle de l'Institut suisse de bioinformatique puisque sa mission et son profil seront différents. L'Institut suisse de bioinformatique s'intéresse aux sciences fondamentales et à toutes les espèces vivantes. Il gère de vastes bases de données, généralement en libre accès dans le monde entier. Le futur centre d'informatique médicale se concentrerait sur la standardisation, la réglementation et les implications éthiques des données numériques sur la santé humaine, sans forcément publier ni stocker des données. Le concept du centre devrait être adapté aux besoins et contraintes des hôpitaux, tels que dictés par les exigences légales, notamment la protection des données, les structures organisationnelles et les systèmes d'information centraux. Par ailleurs, le centre devrait tenir compte de la stratégie et des tâches du bureau de coordination national envisagé par la Conférence suisse des hautes écoles (CSHE) comme infrastructure de base en matière d'information pour toutes les disciplines[69]. Bien sûr, le centre devrait se coordonner avec la stratégie nationale de la Confédération et des cantons et tirer profit de l'expertise accumulée par eHealth Suisse, notamment sur les questions techniques et sémantiques.

Le CSS suggère que le nouveau centre fournisse l'expertise interdisciplinaire nécessaire à la formalisation de concepts de santé et de maladie ainsi qu'à leur traduction en normes sémantiques correspondant au contexte social et aux implications éthiques de l'environnement suisse des soins de santé. Ce centre devra exceller dans des domaines rarement en contact direct tels que l'éthique et les mathématiques. Le centre fera également office de point de contact unique pour les interactions internationales relatives à ce domaine.

La Confédération et les cantons devraient décider d'emblée si le stockage nécessaire à la conservation de données cliniques (voir recommandation 6) devrait être financé via le nouveau centre ou les hôpitaux et/ou les hautes écoles.

9: Le CSS recommande au SEFRI de définir la valeur ajoutée attendue des projets scientifiques, de la collaboration institutionnelle et du partage de pratiques soutenus par l'initiative SPHN, et d'aider à positionner le SPHN au niveau international.

L'initiative SPHN véhiculera les standards à développer et adopter à l'échelle nationale. Une partie de cette mission sera réalisée par un centre national d'informatique médicale, indispensable pour établir le point de contact pour les comités de recherche médicale européens et internationaux. Le SPHN participe déjà aux efforts de coordination tels que le réseau ICPerMed ainsi que la Global Alliance for Genomics and Health, et l'intégration internationale devrait être développée encore davantage au cours de la seconde phase. Il faudrait envisager la manière dont l'initiative suisse pourrait apporter une contribution unique au contexte international. Les activités internationales de la Suisse et ses points forts spécifiques en matière de fiabilité et de densité de réseau (permettant des données de grande qualité) ainsi que les compétences en matière de multilinguisme devraient être pris en compte. Il existe un lien étroit entre les questions de linguistique et de codage informatique, de même qu'une nécessité impérieuse de traduire les concepts informatiques en vocabulaires compris à la fois des patients et des professionnels de santé.

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69   Programme swissuniversities P-5 «Information scientifique».

En fin de compte, le succès de l'initiative SPHN dépendra d'un élément critique, à savoir si les données de patients collectées à large échelle et de manière systématique pourront fournir une valeur ajoutée en matière de qualité des projets de recherche et de qualité des soins dispensés, en comparaison avec les données rassemblées dans le cadre d'essais cliniques spécifiques. Le premier objectif est de garantir l'interopérabilité, comme le stipule la mission de l'initiative pour la période 2017–2020. De plus, il sera intéressant d'étudier l'évolution des pratiques de recherche et la manière dont les institutions collaborent et utilisent les ensembles de données partagés. Enfin, l'initiative pourrait également aider les hôpitaux à développer une stratégie globale d'innovation en tenant compte des caractéristiques uniques du domaine des soins, comme le fait qu'un remplacement extensif des prestataires de soins par l'IA serait probablement vu comme une perte en qualité.

5.2.4
Repenser la formation médicale

10: Le CSS recommande aux autorités fédérales et cantonales de définir un socle de compétences commun pour le bachelor en médecine et de proposer un petit nombre de masters différenciés en médecine.

La pensée conventionnelle veut que les médecins de demain disposent de vastes compétences de base en mathématiques, statistiques, physique, chimie, informatique et génétique ainsi qu'en physiologie humaine. Cependant, la possibilité que l'acquisition de données numériques et l'intelligence artificielle puissent remplacer ou simplifier certaines tâches et compétences (telles que des manipulations techniques et la compilation de connaissances) doit être prise en compte.

La compréhension de méthodes et paradigmes scientifiques ainsi que la maîtrise d'un langage formel – qu'il s'agisse de code informatique, de mathématiques ou de génétique – resteront au cœur de la formation médicale. Les facultés de médecine devraient développer des programmes en informatique clinique qui devraient être envisagés non pas comme une discipline technique, mais comme un effort de sémantique. Tous les médecins seront les «traducteurs» de concepts et notions apportés par les patients via la communication verbale[70] et non verbale, et fourniront à leurs patients la signification d'informations quantitatives provenant de tests in vitro, d'imagerie médicale et de simulations informatiques. Les médecins devront développer de plus en plus des compétences critiques pour évaluer les implications éthiques d'innovations médicales.

La psychologie humaine est un facteur important du processus de guérison parce que les patients attachent une signification personnelle à ce qui leur arrive et parce que certaines interprétations sont plus susceptibles de restaurer la santé que d'autres[71]. Ces compétences requièrent une maîtrise des interactions personnelles qui ne peut être transmise dans un ou deux modules de formation spécialisés. Pourtant, les médecins ne devront pas se contenter de faire preuve d'empathie si, par exemple, ils ne peuvent pas aider leurs patients à comprendre les probabilités.

Le Conseil considère que la maîtrise des sciences et des humanités devrait faire l'objet d'un nouveau programme d'études de bachelor. Il est probable que de nombreux thèmes enseignés actuellement au cours des trois premières années de la formation médicale doivent être déplacés aux programmes de master, qui pourraient être différenciés en un petit nombre de parcours reflétant la diversité croissante des carrières et pratiques professionnelles[72]. Tous les étudiants devraient avoir un premier contact avec la recherche médicale au plus tard durant leur programme de master. Ceci représente une réorganisation d'importance pour faire face aux tendances dominantes de saturation d'informations et recentrer la formation médicale.

11: Le CSS recommande aux autorités fédérales et cantonales de développer de nouveaux programmes de formation dans des disciplines voisines, au-delà du cursus de formation médicale.

On ne peut attendre de la formation médicale qu'elle aborde tous les thèmes liés à la gestion de la santé, à l'ingénierie biomédicale, aux soins infirmiers, etc. C'est pourquoi des programmes interconnectés devraient être mis au point parallèlement à l'évolution des cours médicaux de base. Les activités spécialisées pourraient être déléguées à d'autres professionnels: les consultations génétiques pourraient être assurées par des conseillers en génétique, et les diagnostics simples pourraient être laissés aux infirmiers disposant d'une formation spécialisée. Pour terminer, la médecine devrait être envisagée comme une discipline voisine de l'étude universitaire de la biologie humaine tout en s'en distinguant.

70   S. Hurst (2018). Une histoire dans laquelle vivre. Bulletin des médecins suisses 2018;99(19–20):642.

71   ASSM & CSS (2019). Patient, médecin, big data. Qui a le pouvoir de définition? Swiss Academies Communications. Vol. 14, No 3, 2019.

72   J. Steurer et A. Buchli (2018). Vier Aspekte einer zukunftsfähigen Medizinausbildung. NZZ. 22.03.2018.

Abréviations

ASSH	Académie suisse des sciences humaines et sociales
ASSM	Académie suisse des sciences médicales
CIF	Classification internationale du fonctionnement, du handicap et de la santé
CIM	Classification internationale des maladies
CSHE	Conférence suisse des hautes écoles
CSS	Conseil suisse de la science (1965–1999 et à nouveau depuis 2018) 2000–2013: Conseil suisse de la science et de la technologie (CSST) 2014–2017: Conseil suisse de la science et de l'innovation (CSSI)
DEFR	Département fédéral de l'économie, de la formation et de la recherche
DSM	Manuel diagnostique et statistique des troubles mentaux
EMBL-EBI	European Molecular Biology Laboratory's European Bioinformatics Institute
IA	Intelligence artificielle
ICPerMed	International Consortium for Personalised Medicine
LPGA	Loi fédérale sur la partie générale du droit des assurances sociales
Message FRI	Message du Conseil fédéral relatif à l'encouragement de la formation, de la recherche et de l'innovation
OFSP	Office fédéral de la santé publique
OMS	Organisation mondiale de la Santé
PNR	Programme national de recherche
SEFRI	Secrétariat d'État à la formation, à la recherche et à l'innovation
SNF	Fonds national suisse de la recherche scientifique
SPHN	Swiss Personalized Health Network
SWR	Schweizerischer Wissenschaftsrat (1965–1999 und erneut seit 2018) 2000–2013: Schweizerischer Wissenschafts- und Technologierat (SWTR) 2014–2017: Schweizerischer Wissenschafts- und Innovationsrat (SWIR)

Considerations and recommendations by the SSC

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1

Goal

In its Working Programme 2016–2019, the Council acknowledges a massive increase in the accumulation and availability of health-relevant information. The digitalisation of health care brings far-reaching consequences for all public and private actors. The Swiss Personalized Health Network (SPHN) and other national or international efforts are attempting to regulate and incorporate such data systematically into the practice of medicine and, at the same time, to take the increasing self-monitoring practices of private citizens into account[73]. "Common to these different approaches is the assumption that the "norm" of what is healthy and what is ill is well known and can be defined through measurable parameters, which in turn can be accessed and continuously refined through statistical analysis."[74]

The Council aims to shed light on specific challenges that arise from these new approaches and from the assumptions they are based on, in order to underscore possible consequences for policy recommendations. "Assumptions" are assertions that are accepted as true without proof, for the sake of building an argument. On the one hand, the claim that data sharing allows for medical progress is a verifiable assertion. For instance, children with rare diseases can now hope to receive a timely diagnosis thanks to genomic analysis and/or online platforms where patients share their symptoms around the world, rather than experiencing repeated misdiagnoses at the hands of baffled specialists. Also, the genetic mapping of mutations in roughly 100 oncogenes has proved very useful for defining appropriate treatments with targeted cancer therapies. On the other hand, a number of common assumptions need to be questioned in the context of both research and health care; namely, that data "speak for themselves"; that digital information can be stored and transported at negligible costs or that correlation is an adequate substitute for causation.

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73   For example the Precision Medicine Initiative in the USA, including the cohort All Of Us, the Personalized Medicine Signature Initiative in Canada and the 100,000 Genomes Project in the UK. The database of the International Consortium for Personalised Medicine (ICPerMed) under the link: https://www.icpermed.eu/ offers a more comprehensive view.

74   SSC Working Programme 2016–2019, p. 49. Available under: https://www.wissenschaftsrat.ch/en/programme.

Nowcasting

In 2008, Google Flu Trends was launched, a web service for epidemiological surveillance using search engine query data. Initially, the algorithm appeared to detect influenza outbreaks accurately and faster than established control centres – though it did miss the nonseasonal A–H1N1 pandemic of 2009. In 2015, however, after having repeatedly overestimated the prevalence of seasonal outbreaks, Google Flu Trends was terminated. Expert observers infer that the developers struggled to adapt their model to dynamic patterns of user behaviour, such as local "media hype" phenomena, and to the daily updates of the Google search engine itself[75]. This example highlights a range of issues emblematic for data-intensive research and development: no public access to primary data and limited disclosure about the algorithm, as well as seemingly perfect correlation through recalibration, as both the "measuring lens" and the object under scrutiny are continuously changing.

The setback does not demonstrate that every *nowcasting* application for disease surveillance must fail, particularly when designed as a complement to rather than a substitute of traditional data collection and analysis. Ongoing efforts using various types of web sources could lead to valuable public health advances, especially for developing countries.

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75   D. Lazer et al. (2014). Big data. The parable of Google Flu:
      traps in big data analysis. Science. 2014; 343(6176):1203-5.

This is not a paper in favour or against the digitalisation of health care. As more data and algorithms are being used in the practice of medicine, they are bound to have a profound impact. For their effect to be beneficial, conditions need to be established among the research and health care institutions in terms of data quality standards and modes of collaboration. Moreover, in order to mitigate the trend of medicalisation that is already manifest in high-income countries, there is a need to reflect on expectations with regards to health both on the individual and societal level. The broader context in which big data[76], artificial intelligence (AI)[77] and personalised medicine[78] are implemented, is critically important to the public good. This report presents reflections on the framework conditions of data-intensive approaches in medicine and health care. It is aimed primarily at public authorities in charge of guiding the development of the health care and education systems, which in Switzerland are distributed between the federal and cantonal levels. This report will also contribute to a larger discourse on health that could be of interest to a number of institutions, experts and members of the public, which is of special relevance in a system where wide competences are delegated to civil society actors.

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76   For a definition of 'big data' in the life sciences, see S. Leonelli (2016).
      Data-Centric Biology: A Philosophical Study. The University of Chicago
      press, or S. Leonelli (2017). Biomedical knowledge production in the
      age of big data. Analysis conducted on behalf of the Swiss Science and
      Innovation Council SSIC. Exploratory study 2/2017.

77   AI is the simulation of human intelligence by computer systems and other
      machines. Applications of AI include automated learning, which provides
      systems with the ability to recognise patterns and classify objects such as
      images, sounds or texts, without being explicitly programmed to do so.
      For instance, automated clinical decision support systems analyse patient
      data and help professionals in both diagnosis and prognosis.

78   Through an abundance of data about a patient's overall status and indi-
      vidual characteristics, the goal of personalised medicine is to direct him or
      her to a targeted subset of therapeutic options.

2
Methods

To inform its reflections, the SSC constituted an interdisciplinary working group that undertook a range of discussions and exchanges and reported on these to the Council on various occasions. The concept of health is vast and multidimensional. Obviously, the questions pursued by the Council represent only a selection of all possible aspects and points of view. Perhaps unsurprisingly, the Council's selection remains closely linked to the topics of education, research and innovation, which are at the core of its domain of expertise. Given the prospective outlook of the questions and the rapidly changing institutional landscape, the Council does not have the ambition to reach any final definition of health nor any definitive predictions on the future of health care and health research.

A discursive, cooperative approach was chosen, in dialogue with national and international experts and institutions, to reflect the multiplicity of perspectives, combining workshops with written background analyses (section 3). These expert inputs were further investigated and complemented by literature analyses and exchanges within the working group (section 4). On this basis, the SSC presents its synthesis and recommendations (section 5).

3
Expert inputs

On 15 January 2018, the SSC working group held a seminar in Zurich to hear Ewan Birney, co-director of the European Bioinformatics Institute of the European Molecular Biology Laboratory's European Bioinformatics Institute (EMBL-EBI), and Stephen Senn, Head of the Competence Centre for Methodology and Statistics at the Luxembourg Institute of Health, speak on the scientific potential of big data.

> For Ewan Birney, the enormous amount of data on every person soon to be generated will be a game changer. Research and medical practices will remain guided by different interests: biomedical research has turned into an international endeavour, whereas medical practices are tightly linked to the specific features of local health systems. Supranational boards such as the EMBL-EBI will define common standards and vocabulary for the scientific community. At the national level, centralised organisations should be put in place to coordinate the curation and standardisation of medical data.

> For Stephen Senn, scientists and regulators assume that published data and experimental designs are capable of indicating the scope for personalised medicine, while this is often not the case. A major challenge is to understand the source of statistical variation. A common but unexamined assumption is that genetic factors are more important than other factors of variation, such as patient environment or the inconsistency of medical practice among doctors, or even measurement error. Clever small designs would do more to answer such questions than simply measuring more things on more patients[79].

On 18 April 2018, the SSC working group held a workshop together with the Swiss Academy of Medical Sciences (SAMS). The keynotes speakers were Joachim Buhmann, professor for computer sciences at the ETHZ, and Werner Bartens, medical doctor and editor at the *Süddeutsche Zeitung*. In total, 32 experts participated, and SAMS and SSC reported on the workshop in a joint publication[80].

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79   Also see: S. Senn (2018). Statistical pitfalls of personalized medicine. Nature. 2018 Nov; 563(7733):619-621.

80   ASSM & SSC (2018). Patient, doctor, big data. Who has the power of definition? Swiss Academies Communications. Vol. 14, No 3, 2019.

For Joachim Buhmann, machine learning and AI will have a profound impact on medicine by taking over many tasks and competencies. In the long term, what will probably remain will be physicians' mission as patient advisors. Critical assessment of AI and ethical judgement will ascertain themselves as essential competences for the general public and even more so for medical doctors. The challenge will be to develop strategies in order to evaluate algorithms that are not fully "comprehensible" for human intelligence.

For Werner Bartens, the difference between health and disease is rapidly blurring, to the point that a healthy person is merely someone who has not been diagnosed in sufficient detail. A number of diagnostic methods such as blood tests and computer imaging are neither sensitive nor specific enough to guide sound clinical decisions. Medicine is focused on technical solutions and seemingly exact predictions, to the detriment of skills like communication and empathy that have proved essential to the healing process.

On 30 April 2018, the SSC working group met with Torsten Schwede, chairman of the Scientific Expert Board and director of the Data Coordination Centre of the SPHN, and Christian Lovis, head of the Division of Medical Information Sciences at the University of Geneva and member of the SPHN Scientific Expert Board, to discuss the situation of medical data curation within Switzerland.

For Torsten Schwede, the traditional approach of building large centralised databases is unsuitable for addressing future challenges, due to privacy issues, technical limitations and the dynamic nature of clinical data acquisition and interpretation in modern hospitals. Pooling data and even imposing specific software use is unnecessary. Instead, all actors must acknowledge and share common rules and standards with respect to data annotation and interpretation.

For Christian Lovis, Switzerland could take advantage of its very dense care data landscape to exploit new research avenues. The current data ecosystem of biomedical research is characterised by two aspects: first, the use of "real world data" – acquired in non-controlled environments – implies the need to develop new analytical and validation approaches. Second, cooperation and data sharing call for a common framework of clinical semantic and, at the national level, for the establishment of a dedicated centre (virtual or physical). Importantly, there is a lag in competencies in clinical informatics/phenotype and health semantics in Switzerland, and building capacity in the field and introducing it into existing curricula is urgently needed.

The discussions were enriched and enhanced by expert reports. Sabina Leonelli of the University of Exeter was invited by the Council to analyse the scientific function of big data in the biomedical field[81], and Dominic Murphy of the University of Sydney was asked to discuss the different positions on health and disease as philosophical concepts[82].

For Sabina Leonelli, "data" are never free of some particular scientific context, be it the perspective of the researcher who collected them or the research question of a secondary user. Therefore, the theoretical frameworks of various biomedical subfields and especially their continued scientific evolution must be taken into account when creating and managing databases. Because of the high degree of cross-linking, a single badly curated database may affect data quality on a systemic level. This highlights the need for long-term funding of time-intensive activities such as data management.

For Dominic Murphy, there is no overarching theory of health to serve as a basis for a definition. Overall, the concept of disease has been investigated much more thoroughly than the concept of health. A prevailing understanding of disease, in use in the natural sciences, is that it is a malfunction of nature that can be objectively identified and measured. Other perspectives put the accent on the prior appraisal that a given condition is harmful and therefore identified as a disease, eventually leading to scientific inquiry. The growing use of big data in the sciences and techniques will not shift the dominant understanding of health as a natural object, but could increase the number of conditions considered as pathological.

81   S. Leonelli (2017). Biomedical knowledge production in the age of big data. Analysis conducted on behalf of the Swiss Science and Innovation Council SSIC. Exploratory study 2/2017.

82   See annex to this report.

4
Viewpoints on health

It is well known that there is no consensus notion of health. The World Health Organization (WHO)'s 1948 definition as "a state of complete physical, mental and social well-being" is one of the most famous, but it is also disputed. The counterpart of health, disease, is defined by reference frameworks such as the International Classification of Diseases (ICD)[83] through discrete classes of diseases. In Switzerland, the concept of health is not legally defined, but it is used to circumscribe, among other concepts, disease, namely as "any impairment to the physical, mental or psychological health that is not the consequence of an accident and that requires medical examination or treatment or causes work inability"[84]. How health and disease relate to each other can vary as well. Some conceptions envisage a dichotomy between two mutually exclusive states; others see a continuum between the two ends of a spectrum.

Most perspectives on health focus on one of the following dimensions: freedom from disorders, well-being, functioning, homeostasis, flexibility or capability to adapt[85]. In order to represent the scope of the SSC's reflections in a systematic way, the following section explores four vantage points on health in the context of big data, personalised medicine and AI, addressing the topic as a scientific, ethical, economic and professional issue. The first two perspectives, health as a probability and health as a value, reflect an inner view on the notions of health, while the latter two, health as a market and health as a profession, investigate the implications of such notions of relevance for the Swiss education, research and innovation (ERI) and for the health care systems.

4.1
Health as a probability

Health is increasingly being conveyed through data and perceived as the outcome of computational operations. This poses two kinds of challenges: one pertains to data and computational validity (see 5.1.1) and the second to data interpretation and understanding. With the growing accumulation of high-definition images of the human body, structural anomalies are noted, although one does not know whether they affect health. Full genomes are being decoded at a fast pace, but there is no roadmap to interpret the data at a systemic level. Genetic variants, even for the few that are well understood, do not define "diseases" but a certain level of disease risk[86]. Screening can identify people at risk before they develop a disease. For instance, testing all newborns for rare metabolic conditions prevents a few of them from becoming very sick. It is, however, not always the case that early detection improves survival[87]. Furthermore, new problems can be created in the name of prevention, by overestimating the threats associated with a condition or underestimating those caused by intervention[88]. Finally, whereas a given risk can be computed, no empirical evidence is sufficient to determine the acceptable risk level for a person or a society.

The concept of risk is difficult to grasp for the average patient, and several surveys indicate that not all clinicians are in the position to help him or her understand its meaning. In the words of C. Martyn: "When doctors offer a preventive drug or a screening test to large numbers of asymptomatic people they're doing something quite different from treating a patient who has sought help because she is sick. [...] In this new kind of medicine, not understanding risk is the equivalent of not knowing about the circulation of the blood."[89]

83   The WHO coordinates both the ICD and the International Classification of Functioning, Disability and Health (ICF). Another example of reference framework is the Diagnostic and Statistical Manual of Mental Disorders (DSM) published by the American Psychiatric Association.

84   The Federal law on the general part of the law of social insurance delimitates the topic of health within discrete sectors (accidents, disability, disease) governed by separate laws: "Est réputée maladie toute atteinte à la santé physique, mentale ou psychique qui n'est pas due à un accident et qui exige un examen ou un traitement médical ou provoque une incapacité de travail" (Loi fédérale sur la partie générale du droit des assurances sociales LPGA, art. 3 al. 1).

85   A. Franke (2012). Modelle von Gesundheit und Krankheit. Dritte überarbeitete Auflage. Bern: Verlag Hans Huber.

86   See annex to this report.

87   H. G. Welch (2018). The heterogeneity of cancer. Breast Cancer Research and Treatment. Vol. 169, Issue 2, pp. 207–208.

88   See for instance: US Preventive Services Task Force (2017). Hormone Therapy for the Primary Prevention of Chronic Conditions in Postmenopausal Women. JAMA. 2017; 318(22):2224–2233.

89   C. Martyn (2014). Risky business: doctors' understanding of statistics. BMJ. 2014; 349:g5619.

Predicting autism

Autism is a developmental disorder affecting, according to recent estimates, 17 out of every 1000 US children. Its causes are not well understood; the predisposing genetic indicators are broad, and detection at a young age is very challenging. By 2 years of age, autistic children display readily identifiable social withdrawal behaviour patterns. In 2017, a US team reported successful diagnosis of autism in 6 months-old infants with 81% accuracy and 88% sensitivity with the help of brain scans and deep-learning[90]. Soon afterwards, even better performances were being reported by analysing electroencephalogram patterns in 3 months-old infants, reaching an accuracy and sensitivity of 95% each[91].

An accuracy of 95% appears vastly superior to currently established methods. Still, the proportion of false positive results argues for only proposing the test to infants already considered to be at risk: knowing nothing about a child apart from his or her positive test result means that the probability for the child to be autistic by age 2 would be only 25%[92] – and no more than 7% in the less accurate brain scan test. It is likely that early intervention attenuates later symptoms, thus bringing a very significant benefit to affected children, while an incorrect positive result probably causes some harm to a child's development and relationship with his family. In a best-case scenario where early treatment effectively helps preventing social difficulties, it might even become impossible to discriminate between "true" and "false" positives later on, also because autism is viewed as a gradual and not a discrete condition.

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90   H. C. Hazlett et al. (2017). Early brain development in infants at high risk for autism spectrum disorder. Nature 2017; 542(7641):348-351.

91   W. J. Bosl et al. (2018). Early brain development in infants at high risk for autism spectrum disorder. Sci Rep. 2018 1; 8(1):6828.

92   Of 1000 children, 934 will correctly receive a negative diagnostic, 16 will be autistic with a correct positive diagnostic, 1 will be autistic but falsely labelled as negative and 49 will be false positives.

4.2
Health as a value

Based on how much Swiss citizens are ready to invest, both in public and private spending,[93] it would appear that health – and security in general – is viewed as the highest good. Yet, the behaviour of many individuals indicates that they consider their health as a relative good, for example prioritising "work ethics" or the freedom to not always make the "right choice" concerning their health. These values and their ethical ramifications shape the Swiss system of social insurances. Health, after all, is also a public good, in the sense that even the richest and healthiest people gain in security if other members of society suffer from fewer infectious diseases, mental disorders or addictions[94].

Today, a growing trend is to consider health as being primarily shaped by individual behaviour. Both the personalised medicine approach and the *quantified self* movement focus on the individual as opposed to society, as the source of data accumulation and refining predictions. The hope for technology to empower individuals ultimately calls into question society's overall disposition to solidarity[95]. In view of the general availability and ubiquity of data, a major ethical issue for the conflicting priorities of solidarity and autonomy will be whether today's "right not to know" (see next page) can be preserved in the future.

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93   In 2016, overall health care spending reached 80.7 billion CHF or 12% of the Swiss Gross Domestic Product. In 1996, the same item received 37.5 billion CHF or 9% of the national GDP.

94   Furthermore, an economic argument could be made in terms of welfare payment increase and tax payment loss.

95   F. Gutzwiller & A. Müller (2017). Gerechtigkeit im digitalen Zeitalter. Personalisierte Daten – eine Frage der Solidarität. NZZ. 18.10.2017.

The right not to know

A large proportion of patients at risk of developing Huntington's disease prefer not to undergo genetic testing[96]. Clearly, knowledge of an impending disease entails psychological costs susceptible to impact quality of life years before outbreak. Furthermore, even for carriers of a dominant autosomal mutation such as Huntington, the clinical outcome's prediction remains imprecise as the severity, age of onset and speed of progression vary widely. In any case, the decision for or against genetic testing is never taken based on strictly personal factors: children, siblings and even distant cousins will probably be impacted by the outcome of a test they did not ask for.

Different countries vary in their conception of family solidarity obligation. According to French law, a person who learns of a genetic risk has the duty to inform her relatives, although the patient can ask the attending doctor to do it on his or her behalf (*Loi relative à la bioéthique du 7 juillet 2011*, art. 2). In Switzerland, the same person is advised to disclose the information, without being liable if she prefers not to. In this case, however, the attending doctor must decide whether the interest of the patient's relatives may outweigh confidentiality obligations towards the patient.

Out of respect for personal autonomy, article 8 of the Federal Act on Human Genetic Testing (newly revised in June 2018) states that "[e]very person has the right to refuse to receive information about his or her genetic status, completely or partially". The "right not to know" is the expression of an active and explicit wish of the concerned person. Circumstances where it applies may become more frequent, including situations beyond genetic counselling, given the growing number of predictive algorithms. Of course, its practicability remains limited, as a patient must know that he or she is at risk before she can decide against receiving the information.

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96   P. J. Morrison (2010). Accurate prevalence and uptake of testing
      for Huntington's disease. Lancet Neurol. 2010; 9(12):1147.

4.3
Health as a market

Improving health is the one goal for which people find it difficult to cap their investments. Therefore, health care is the ultimate sector of growth, offering jobs and export opportunities, and placing public finances in front of ever more challenging cost increases[97]. Unique characteristics make health care a distorted market, such as wrong incentives and knowledge imbalance between providers and consumers[98]. Physicians often choose less medical care for themselves than they recommend to patients[99]. They are, arguably, in the best position to realise that more treatment does not automatically mean better outcomes, while the current system is rewarding treatment, not outcomes. In Swiss hospitals, yearly bonuses for reaching minimal numbers of interventions are common. Many patients, as a result, especially if they are privately insured, are encouraged to undergo unnecessary procedures, thus risking adverse events and side-effects[100]. Doctors in private practice are better rewarded for technical interventions than for intellectual and communicative tasks.

There is a clear political pressure to lower the costs of the disability insurance system, with a focus on promoting rehabilitation and making it increasingly difficult for new applicants to be granted a disability allowance. The pressure is somewhat weaker in the general health insurance system, because voters generally reject proposals that restrict their range of care options. Several conflicts of interests make systemic changes very difficult, be it at the level of citizens (see for example: initiative on single insurer, referendum on managed care, local decisions related to closing hospitals), members of parliament (votes on

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97   See note 93 p. 35.

98   OFSP (2017). Mesures visant à freiner la hausse des coûts dans l'AOS.
      Rapport du groupe d'experts du 24.08.2017.

99   K. Murray (2011). How Doctors Die. Zócalo Available under:
      http://www.zocalopublicsquare.org/2011/11/30/how-doctors-die/
      ideas/nexus/; V. S. Periyakoil et al. (2014). Do Unto Others: Doctors'
      Personal End-of-Life Resuscitation Preferences and Their Attitudes
      toward Advance Directives. PLOS.

100   O. Peters et al. (2016). Excédent de prestations stationnaires pour les
      personnes avec une assurance complémentaire. OFSP 10.06.2016;
      F. Gottschalk et al. (2017). Health Services as Credence Goods: A Field
      Experiment. ETH Zurich, September 2017.

prevention, tobacco regulations) and health care providers (TARMED tariffs negotiations)[101]. If current trends continue, the costs for health care may rise to a point where they actually make the Swiss population less healthy by constraining discretionary spending for other public tasks, especially at the cantonal level.

4.4
Health as a profession

Medical doctors are central to the health care system. It is their responsibility to put a name to a condition and to assess whether someone is sick, and as such, entitled to benefits such as treatment or rest from work. For instance, mandated by the disability insurance, medical experts define whether persons – who are not their patient – are unable to work, a difficult task in cases of chronic pain or mental disease. In a typical doctor's practice, however, the problem is not the lack of measurable "evidence", but the absence of straightforward formula to extract knowledge from it. Given the speed at which gene sequences and three-dimensional images are generated, it is often unclear which kind of doctor, the general practitioner or one of the various specialists, should interpret which piece of information[102]. Even for common laboratory tests and common diseases, data semantics and symptoms descriptions are not uniformly used across hospitals[103]. Today's patients often arrive at a doctor's office bringing their own hypotheses about their health. Finally, each patient has an individual opinion on his or her goal in terms of health, which cannot be assumed to be aligned with the doctor's goal.

A general assumption is that tomorrow's doctors will need an even broader range of competence than today: more physics and biochemistry, better social skills and communication, a deeper mastery of probabilities, some notions on alternative medicine and possibly knowledge of medical informatics, coding and engineering. The result could be that medical education is on the brink of overload, which may lead, despite best efforts, to a focus on accumulating facts and teaching to the test.

Other professionals with essential roles for the health care system, such as nurses, are abandoning their learned profession in high proportion – thereby demonstrating that attracting and forming more candidates is not sufficient to ensure adequate numbers of caregivers[104]. There are reasons to believe that, had working conditions been better, many of those caregivers would have continued working[105]. Swiss citizens will vote on a popular initiative proposing, among other things, to redistribute some competences between physicians and nurses[106].

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101 Ongoing political discussions aiming to harmonise payments modes could, if successful, remove the incentive for hospitals to perform inpatient rather than outpatient treatments. This measure, however, would not shift the prevalent prioritising of quantity over quality of treatment.

102 R. R. Stavert & J. P. Lott (2013). The Bystander Effect in Medical Care. NEJM. 2013; 368:8–9.

103 A. K. Manrai, C. J. Patel & J. P. A. Ioannidis (2018). In the era of precision medicine and big data, who is normal? JAMA. 15 May 2018, Volume 319, Number 1.

104 In 2013, 32% of all nurses 35 years and younger had left their learned profession. This proportion rises to 56% for nurses 50 years and older. (M. Lobsiger et al. [2016]. Les professionnels de la santé: sorties de leur profession. Obsan Bulletin 7/2016, Neuchâtel.)

105 R. Schwendimann et al. (2014). Das Pflegefachpersonal in Schweizer Spitälern im europäischen Vergleich. Obsan Bulletin 3/2014, Neuchâtel; C. Merçay et al. (2016). Personnel de santé en Suisse. Etat des lieux et projections à l'horizon 2030. Obsan Rapport 71, Neuchâtel.

106 https://www.bk.admin.ch/ch/f/pore/vi/vis472t.html.

5
Position of the SSC

5.1
Synthesis

Based on external inputs and internal perspectives presented in the previous sections, the Council proposes action in the fields of research and innovation, education and health promotion. The SSC understands its position as being complementary to other voices that bring important contributions on topics such as cost containment[107], data governance[108], ethics[109] or quality of life[110], to name a few.

5.1.1
Research and innovation

Analytic approaches that exploit large datasets open up a range of new possibilities for science and medicine. Their potential is evident in the development of new diagnostics, and progress is ongoing to support decision making in the context of therapeutic options. These approaches can also lead to a better understanding of pathological mechanisms through the identification of unsuspected correlations. Yet in order to establish causal relations, experimental confirmation remains indispensable. Given the propensity of experts and non-experts alike to perceive expensive interventions as superior to cheaper alternatives[111], health care innovations should demonstrate superiority based on clinical evidence.

Of scientific importance are issues related to (i) the quality of data processing during collection, annotation and storage, (ii) the publication of so-called negative research results and (iii) the reproducibility of published experiments. While these challenges are in no way specific to large datasets, the problems

are unlikely to be alleviated without explicitly addressing them. Aggregating large numbers of biased datasets does not ensure equitable representation and rather leads to reproducibility challenges because of technical limitations, lack of access to data and to algorithms[112]. These concerns for scientific validity may explain why a large number of researchers use only data of colleagues they know personally. The successful sharing of large datasets requires that one defines and ensures implementation of semantic terminologies and processing standards. A national coordinating site or institute is necessary to evaluate and enforce the best standards as they are in continual scientific evolution.

5.1.2
Education

Large digital datasets are a prerequisite for machine learning and AI, and both will transform the practice of medicine. This also changes the skills needed by future health care professionals. Important are skills in synthetic thinking and logical deduction, while encyclopedic knowledge or fact accumulation are increasingly delegated to support systems. Indeed, machines are taking over a growing number of technical tests and acts currently performed by physicians. Future medical doctors will need to develop advanced competence in the critical assessment of AI and will need to master the language of algorithms as well as patients' narratives and non-verbal communication. Above all, they will have to sympathise with a patient's unique perspective in order to help him or her navigate the growing complexity of care. These insights are calling for changes in medical education.

5.1.3
Health promotion

Both the philosophical analysis[113] and the expert discussion[114] agree that large digital datasets cannot free the concept of health of its dual subjective and objective nature. No socially neutral or value-free notion of health is possible. Therefore, science and medicine need to acknowledge that ethical and social concerns influence the choice of data and meta-data that health care professionals will monitor. Overall, the data wave will present the health care system with two possible outcomes. It can lead to a further expansion of the common understanding of what is pathological, converting more and more people into at-risk individuals and nearly every condition into

107   OFSP (2017). Mesures visant à freiner la hausse des coûts dans l'AOS. Rapport du groupe d'experts du 24.08.2017.

108   B. Prainsack (2015). Personalized medicine. Empowered patients in the 21st century? New York City: New York University Press: Biopolitics December 2017.

109   European Commission Science Advice and Ethics Groups at ESOF (2016). Ethics of New Health Technologies and Citizen Participation. Opinion n° 29 – 13/10/2015; European Group on Ethics in Science and New Technologies (2018). Statement on the Ethics of Artificial Intelligence, Robotics and "Autonomous Systems"; Ethical Framework for Responsible Data Processing within SPHN (Version 1, 12.06.2017).

110   SAHS (2016). Définir, mesurer et promouvoir la qualité de vie. Lebensqualitätsforschung in der Schweiz.

111   R. L. Waber et al. (2008). Commercial features of placebo and therapeutic efficacy. JAMA. 299, 1016–1017; A. J. Espay (2015). Placebo effect of medication cost in Parkinson disease: a randomized double-blind study. Neurology 2015 Feb. 24; 84(8):794–802.

112   J. T. Wilbank & E. J. Topol (2016). Stop the privatization of health data. Nature. Vol. 535, July 2016.

113   See annex to this report.

114   ASSM & SSC (2019). Patient, médecin, big data. Qui a le pouvoir de définition? Swiss Academies Communications. Vol. 14, No 3, 2019.

a rare disease[115]. Alternatively, it could promote the notion that multiple modes of being healthy coexist. Instead of focusing only on the diversity of pathological risks, science could investigate how much variation there is between two healthy individuals, in order to advance the concept that being outside of a physiological or psychological norm is not automatically equated to being ill.

5.2
SSC recommendations and rationale

5.2.1
Promote a comprehensive understanding of health

1:    The SSC invites the Federal Council to prioritise investments in basic education, social programmes and environment preservation as an integral part of health-promoting measures.

Because health is not a purely objective notion, some approximations are necessary to speak quantitatively of health at a population level; life expectancy is one of such indicators. Adopting average life expectancy as a "health proxy" does not imply that extending life and improving health will always be synonymous, but that such statistical information can be a source of learning about the conditions most conducive to health[116]. Studies on longevity have shown that health care is by far not the only parameter contributing to health.

Why do we live longer?

The period following the Second World War was a time of justified optimism about using science to solve major health issues, especially communicable diseases such as tuberculosis, polio or measles. Thomas McKeown (1912–1988), a British medical doctor, questioned the intuitive relationship between longevity and health care, pointing to economic growth, rising living standards and improved nutrition as the main agents of change. He observed that the bulk of mortality reduction of most infectious diseases happened prior to the discovery and deployment of antibiotics. McKeown's assertions have been disputed later on. Today, the following factors are considered by the WHO to explain why a child born in 2018 is likely to reach old age in most countries: nutrition, sanitation, maternal educational level as well as access to medical technologies such as vaccines and antibiotics. Scientific advances underlie nearly all these aspects, yet health care per se is only one part of the equation.

Socioeconomic status continues to correlate with longevity, even in high-income countries. In Switzerland, the difference in life expectancy between inhabitants of rich versus poor city neighbourhoods is 4.5 years for men and 2.5 years for women[117]. In the UK, the corresponding gap reaches 9 years for men and 7 years for women[118].

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117   FOPH (2018). Health equity – Facts and figures for Switzerland.

118   Public Health England (2017). Reducing health inequalities: system, scale and sustainability.

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115   See annex. See also R. H. Moynihan et al. (2013). Expanding Disease Definitions in Guidelines and Expert Panel Ties to Industry: A Cross-sectional Study of Common Conditions in the United States. PLoS Med 10(8): e1001500.

116   This is especially the case when declining life expectancy trends are being investigated. See for instance: S. H. Woolf & L. Y. Aron (2013). The US health disadvantage relative to other high-income countries: findings from a National Research Council; Institute of Medicine report. JAMA. 2013; 309:771; S. H. Woolf et al. (2018). Changes in midlife death rates across racial and ethnic groups in the United States: systematic analysis of vital statistics. BMJ. 2018; 362:k3096.

Several models account for the diversity of health determinants, and more research is needed in order to understand their relative contributions[119]. However, a consensus of the last decade is to consider structural determinants – factors that cannot easily be changed by individuals, such as habitat and work conditions – as more important than behavioural factors, such as diet or physical exercise[120]. Without denying the hope that individuals can increasingly take advantage of knowledge and technology to improve their own health, it seems clear that altering behaviour is not sufficient and that there is a need for policies that encompass social protection, fiscal policy, workplace regulation and environment, transport security and many more sectors. Especially, a high-quality education for every individual is widely recognised as one of the most significant factors of health[121]. All efforts should be directed towards ensuring that public authorities do not find themselves forced to cut their investment in basic education due to short-sighted budget issues.

Furthermore, effective preventive programmes should be developed, not in the – often misguided – expectation that they would reduce costs, but based on the knowledge of their high potential for improving health[122]. Preventive programmes encompass a great variety of measures. Taking the example of obesity prevention, R. Z. Goetzel proposes: "Meaningful changes in social norms can be achieved through coordinated efforts involving multiple stakeholders (for example, urban planners, food growers and distributors, educators, government officials, civic leaders, and health care professionals) without relying exclusively on expensive medical personnel for improving health."[123]

2: **The SSC recommends to the FOPH and the SERI to ensure long-term support of health services research.**

It should be noted that "large" datasets mean different scales at different times[124]. In that sense, scientists have been grappling with large and complex datasets for decades, including in fields such as epidemiology and public health. At present, "big data" in the biomedical context usually means a combination of genomics, proteomics or metabolomics. Mechanisms should be reinforced and new ones introduced to allow the fruitful integration of such molecular approaches with other forms of research. Different kinds of information such as insurance claims, research cohorts or environmental studies, as well as results from epidemiological studies, need to be integrated into health evaluation algorithms and processes. Of help will be data-intensive methods that allow the tracking and observation of voluntary subjects in the "real world", made easier by new technological devices.

Health services research – understood as the scientific field aimed at identifying effective ways to organise, finance and deliver high-quality care – was established some 40 years ago in the United States. This kind of scientific knowledge is crucial in order to establish when and where "less is more" in a careful and differentiated manner[125]. In Switzerland, first steps have been taken with initial funding by the SAMS and the Gottfried und Julia Bangerter-Rhyner-Foundation (2012–2016), later on through the National Research Program (NRP) 74 "Smarter healthcare" (2017–2022). The support will be needed even after completion of the NRP, as this research field must continue to grow. So far, notably, a large number of studies in health services research are describing variations in quality and efficiency. Providing theories of causation and inferential models would greatly enhance the value of this research in remediating problems.

3: **The SSC recommends to the SERI and the FOPH to encourage a continuous debate to explore society's expectations with respect to health care and quality of life.**

A complex integration of many parameters goes into the definition of health. The growing range of diagnostic options available could end up delineating a very narrow standard, reinforcing the trend towards exaggerated medical treatment (called medicalisation) already found in high-income societies. Public authorities are already aware of the importance of communication within the health care sector. Building on existing private initiatives,[126] they should promote the development of new platforms of exchange across disciplines and fields and between the public and the researchers, challenging the notion that health care should prolong life at all costs.

119 N. Cantoreggi (2010). Pondération des déterminants de la santé en Suisse: Etude réalisée dans le cadre de l'élaboration d'un modèle de déterminants de la santé pour la Suisse. Université de Genève.

120 M. Richter et al. (2011). Determinanten der Gesundheit und ihre relative Bedeutung für die Erklärung gesundheitlicher Ungleichheiten: Ein systematisches Review. Universität Bern.

121 FOPH (2018). Health equity – Facts and figures for Switzerland.

122 A. E. Carroll (2018). Preventive Care Saves Money? Sorry, It's Too Good to Be True. The New York Times, 29.01.2018.

123 R. Z. Goetzel (2009). Do Prevention Or Treatment Services Save Money? The Wrong Debate. Health Affairs Vol. 28, No. 1: The Crisis In Chronic Disease.

124 S. Leonelli (2017). Biomedical knowledge production in the age of big data. Analysis conducted on behalf of the Swiss Science and Innovation Council SSIC. Exploratory study 2/2017.

125 A. Gawande (2015). Overkill. The New Yorker, 11.05.2015.

126 TA SWISS, Science et Cité, the Leenards Foundation with SantéPerSo and miDATA to name just a few such initiatives.

Among medical doctors, movements such as the "Choosing wisely" association have brought to prominence discussions on the costs and benefits of medical interventions. These exchanges often lead to uncomfortable but important debates on limitations and valuation of human life[127] and also about the limitations of medicine. To include perspectives from natural sciences, engineering, social sciences and humanities is a necessary, but insufficient step to come to a consensus on health. Health care professionals, patients and private citizens should be included in a debate on the goal of health care in a larger social context[128], to outline what Switzerland, as a society, understands as health, and to determine the degree to which quantitative data analysis should be definitive.

5.2.2
Develop data standards conducive to health

4: The SSC recommends to the FOPH to reduce publication bias in the clinical literature by ensuring access to the results of clinical trials.

Large-scale data mining relies on scientific literature and on open-source databases in order to gain the broadest possible basis for evaluation. However, it appears that experiments yielding expected results are far more likely to get reported and published. Thanks to regulations that require registration of all clinical trials at their onset (in Switzerland since 2014 with the Human Research Act), it has become possible to calculate what proportion of studies are completed and published, a percentage that remains as low as one out of two trials becoming published, with high variation among countries and types of sponsors[129]. One reason for not reporting the results of a trial may be that it was abandoned due to poor feasibility. In other cases, however, the scientist may choose not to publish because the results did not support the initial hypothesis. Human subjects who enroll in clinical trials should expect their participation to help advance science regardless of the private interests of research promoters. Even studies that are interrupted due to insufficient patient recruitment or for other technical reasons

should be reported – albeit not as a traditional publication – to save future clinical projects from repeating past failures. The Confederation should encourage reporting the results of all clinical trials by raising awareness to this issue at the international level, by ensuring accurate reporting of metadata of said trials and by supporting databases and/or new publication formats for the reporting of negative outcomes.

5: The SSC recommends to the FOPH to increase expertise and define validation criteria for medical software.

While the publication bias in scientific literature is recognised, the issue of clinical effectiveness and social repercussions of AI in the health care industry is relatively new. According to the Therapeutic products Act, certain apps, autonomous web-based instruments and softwares embedded in surgery robots or CT scans are medical devices that should be controlled by bodies of accreditation through a simplified procedure. Today's algorithms, however, are often too complex to be understood by regulators and users. It is unclear whether data generated by new technical devices are legally protected, and it is increasingly difficult to define a system's specifications or to evaluate the quality of training datasets used by machine learning algorithms. Updates may represent an inconsequent change or one with far-reaching repercussions, yet they are rarely subject to control. Today's regulatory system assumes that hospitals and medical doctors are sufficiently informed about the algorithms their instruments use, yet this needs to be verified. Increased expertise and better criteria are needed in order to refine the evaluation of medical software, both at national and international levels, from the vantage point of efficacy, technical performance, and with respect to the software's social implications[130]. The new centre that the SSC suggests to establish (see recommendation 8) could play a leading role in this matter.

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127 D. Friedli (2018). Ein Leben ist dem Bund plötzlich fast doppelt so viel wert. NZZ am Sonntag. 07.07.2018.

128 On the goals of health care, see the forthcoming SAMS Roadmap (2019). Sustainable development of the health care system; On public participation, see B. Strasser & M. Haklay (2018). Citizen Science: Expertise, Democracy and Public Participation. SSC policy analysis 1/2018.

129 B. Goldacre (2018). Compliance with requirement to report results on the EU Clinical Trials Register: cohort study and web resource. BMJ. 2018, 362:k3218; A. Amstutz et al. (2018). Funding characteristics of randomised clinical trials supported by the Swiss National Science Foundation: a retrospective cohort study. Swiss Med Wkly. 2018; 148. w14587.

130 See the example of IBM Watson for a discussion of cultural bias in medical softwares: C. Ross & I. Swetlitz (2017). IBM pitched its Watson supercomputer as a revolution in cancer care. It's nowhere close. STAT 2018.

6:   The SSC recommends that the SERI and the cantonal authorities support critical data services such as annotation and curation.

Securing sustainable funding for the essential task of annotating and curating data is difficult. Even the activities of the Swiss Institute of Bioinformatics with respect to its most-used resource (e.g., UniProt) depend largely on American funding sources. The SNSF does not consider data annotation, apart from exceptional cases, as being part of direct project costs. Novel models for distributed funding through research agencies could be investigated, particularly for basic life sciences[131]. With respect to clinical data, however, there is a clear need for sustainable funding and for the enforcement of annotation standards, which is arguably the responsibility of local public authorities. One solution would be to encourage Swiss hospitals and/or higher education institutions to create and maintain this service. An alternative would be to integrate it into a national centre for medical informatics (see recommendation 8). Local database curation following recommendations from a central organ would be the ideal solution.

7:   The SSC recommends that the FOPH and the SERI ensure that the social implications of data choice and specification are taken into account.

T. Wilbanks and E. Topol stated in 2016: "If undisclosed algorithmic decision-making starts to incorporate health data, the ability of black-box calculations to accentuate pre-existing biases in society could greatly increase. [...] It is not hard to picture a future in which companies are able to trade people's disease profiles, unbeknown to the patients. Or one in which health decisions are abstruse and difficult to challenge, and advances in understanding are used to aggressively market health-related services to people – regardless of whether those services actually benefit their health." Predictive algorithms should be used and distributed with great care and must be evaluated by independent experts, fluent in ethics and informatics.

Only few health-related algorithms are actually in use in the Swiss health care system. One could, however, imagine the following uses:

—— Decision-making systems for prioritising patients for organ allocation. Transplantation medicine is already making use of an "algorithm" or fixed set of rules at the national level. Using AI to refine this algorithm would be the next step.

—— Assessment whether or not someone is living a healthy lifestyle: insurance companies are already using "algorithms" to decide whether someone can have private insurance.

—— Predictive models to connect high-need residents to local social services: such public health initiatives could be impaired by a lack of representative data, as poor people tend to generate less data.

—— Detection and forecasting of infection dynamics such as HIV, Ebola, specific flu strains: epidemiology could refine its models using AI. False positive predictions, however, may generate economic stress on cities and countries.

—— Recently, scientists at Stanford University developed a software to estimate how much longer a person is likely to live and when to speak with him or her about palliative care.[132] Care should be taken to ensure that such an algorithm does not contribute to widening inequalities.

If no attention is given to the social impact of such algorithms, minorities with regard to age, disease, ethnic and sociological characteristics are at risk of both under- and overtreatment. Furthermore, the experts who set the thresholds between "normal" and "pathological", which data to collect, how to annotate it and how to share results for determining clinical ontology exert a critical responsibility towards defining health. Their independence from all other actors is very important.

131   C. Gabella et al. (2017). Funding knowledgebases: Towards a sustainable funding model for the UniProt use case. F1000Research 2018, 6 (ELIXIR):2051.

132   A. T. Hsu (2018). Algorithm for predicting death among older adults in the home care setting: study protocol for the Risk Evaluation for Support: Predictions for Elder-life in the Community Tool (RESPECT). BMJ Open. 2016; 6(12):e013666.

5.2.3

Precisely define the goals of the SPHN initiative

8: The SSC recommends that the SERI, the FOPH and the cantonal authorities establish a national centre for medical informatics and medical data analysis, which will set standards and provide guidance to the Swiss medical establishment within the context of the SPHN initiative.

The quest for novel scientific insights based on large sets of data has been decisive in the implementation of major research endeavours, comprising a wide range of scientific interests, from the Human Genome Project (1990–2003) to the Human Brain Project (2013–2022) as well as national initiatives such as SystemsX.ch (2008–2016) and the Swiss Personalized Health Network (started 2017). Large research projects can be considered a crystallisation point for understanding dominant biomedical concepts and for reflecting on their possible medical impact. The SPHN, for its part, is tasked with building bridges across institutions, disciplines and researchers in order to ensure health data interoperability.

Experience suggests that, in the longer term, such a mission requires a coordinating structure. Therefore, the next phase of SPHN should continue its current efforts to build up and support competences at the national level. During the second phase of SPHN, a national centre for medical informatics should be established with a sustainable funding consistent with its essential functions, which should focus on the setting of standards and evaluation of algorithms to be implemented in medical institutes across Switzerland.

In the long term, a Switzerland-wide medical informatics centre should be organisationally distinct from the Swiss Institute of Bioinformatics, as it would have a different mission and profile. The SIB is dedicated to basic life sciences and all species. It is taking care of large databases, usually available for anybody in the world to access. The future centre for medical informatics would be concerned with standardisation, regulation and ethical implications of human digital health data, and would not necessarily publish nor store data. The concept of the centre should be adapted to the needs and constraints of hospitals as defined by their organisation structures and central information systems, and by the law, especially data protection issues. Furthermore, it should take into account the strategy and tasks of the national coordination office envisioned by the Swiss Conference of Higher Education Institutions (SCHEI) as a basic infrastructure for all disciplines in the realm of scientific information[133]. Of course, the centre should also coordinate with the national strategy of the Confederation and the cantons and benefit from the expertise of eHealth Suisse, especially in semantic and technical issues.

The CSS suggests that the new centre could provide the interdisciplinary expertise necessary for the formalisation of concepts of health and disease, and their translation into semantic standards that match the social context and ethical implications of the Swiss health care environment. This will require excellence in domains that are rarely in direct contact, such as ethics and mathematics. The centre will also provide a single contact site for international interactions relevant to this domain.

Whether the data storage necessary to curate clinical data (see recommendation 6) should be funded through the new centre or through the hospitals and/or higher education institutions should be decided early on by the Confederation and the cantons.

9: The SSC recommends that the SERI defines the added value expected for scientific projects, institutional collaboration and sharing practices promoted by the SPHN initiative, and helps position the SPHN initiative internationally.

The SPHN initiative will be a vehicle for data standards to be developed and adopted at a national level. Part of this will be achieved through a national Medical informatics centre, necessary to establish a contact point for European and global medical research boards. The SPHN is already integrated in coordination efforts such as the ICPerMed network and the Global Alliance for Genomics and Health, and international integration should be further developed in the second phase. It should be explored how the Swiss initiative can make a unique contribution in the international context. The international activities of Switzerland and its specific strengths in network density and reliability (making high-quality data possible) and multilingualism should be taken into account. There is a deep connection between languages and computer coding, and a strong need to translate informatics concepts into vocabularies that both patients and health care professionals understand.

Ultimately, the success of the SPHN initiative will depend on one critical point: whether patient data collected systematically and more broadly bring qualitative added value to both research projects and care, as compared to data gathered within specific clinical trials. The first goal is to ensure interoperability, as stated in the mission of the initiative for the period 2017–2020. In addition, it will be of interest to explore how research practices evolve and how institutions will collaborate and make use of shared datasets. Finally, the initiative could also help hospitals to develop a global strategy for health care innovation, taking into account the unique characteristics of health care, such as the fact that a thorough replacement of caregivers with AI would likely be deemed a loss in quality of care.

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133 swissuniversities programme P-5 "Scientific information".

5.2.4

Rethink medical education

10: The Council recommends to the federal and cantonal authorities to define a common core of competencies to be covered by the Bachelor in Medicine and to propose a small number of differentiated Masters in Medicine.

Conventional wisdom is that future medical doctors will require broad basic competence in maths, statistics, physics, chemistry, computational science, and genetics as well as human physiology. However, the possibility that digital data acquisition and AI might replace or simplify certain tasks and skills (such as technical manipulations and the compilation of knowledge) should be taken into account.

Understanding of scientific methods and paradigms and mastery of a formal language – be it computer coding, mathematics or genetics – will remain a central component of medical education. Medical faculties should develop curricula in clinical informatics, which should be understood not like a technical discipline but largely as an endeavour in semantics. Importantly, all physicians will be the "translators" of concepts and notions that patients bring along through narratives[134] and non-verbal communication, and they will convey back to their patients the meaning of quantitative information from in-vitro tests, medical imaging and computer simulations. Increasingly, medical doctors will need to develop critical skills to appraise the ethical implications of medical innovations.

Human psychology is important to the healing process because patients attach personal meaning to what is happening to them, and because some interpretations are more conducive to health than others[135]. Such skills require a mastery of personal interactions that cannot be transmitted in one or two specialised teaching modules. Yet, it will not be sufficient for medical doctors to show empathy, if, for example, they are unable to help their patients comprehend probabilities.

The Council understands that a mastery of both science and humanities topics would be the subject of a new Bachelor study course. It is likely that many subjects currently taught during the first three years of medical education would need to be postponed to the Master programmes, which could be differentiated into a small number of tracks, reflecting the growing diversity of professional careers and practices[136]. All students should get a first contact with medical research during the course of their Master programme at the latest. This would represent a significant reorganisation intended to counter powerful trends of information overload and to refocus medical education.

11: The Council recommends that the federal and cantonal authorities develop new curricula at the boundaries to other disciplines beyond the medical track.

Medical education cannot be expected to address all issues related to health management, biomedical engineering, nursing, etc. Therefore, interconnected curricula should be developed in parallel to the evolution of the core medical courses. Expert activities could be delegated to other professionals: genetic consultations could be given by genetic counsellors, and simple diagnostics could be performed by nurses with specialised training. Finally, medicine should be understood as related to, but distinct from the academic study of human biology.

134 S. Hurst (2018). Une histoire dans laquelle vivre. Bulletin des médecins suisses 2018; 99(19–20):642.

135 ASSM & CSS (2019). Patient, médecin, big data. Qui a le pouvoir de définition? Swiss Academies Communications. Vol. 14, No 3, 2019.

136 J. Steurer & A. Buchli (2018). Vier Aspekte einer zukunftsfähigen Medizinausbildung. NZZ. 22.03.2018.

Abbreviations

AI	Artificial intelligence
ATSG	Bundesgesetz über den Allgemeinen Teil des Sozialversicherungsrechts
DSM	Diagnostic and Statistical Manual of Mental Disorders
EMBL-EBI	European Molecular Biology Laboratory's European Bioinformatics Institute
EAER	Federal Department of Economic Affairs, Education and Research
ERI-Dispatch	Federal Council Dispatch on the Promotion of Education, Research and Innovation
FOPH	Federal Office of Public Health
ICD	International Classification of Diseases
ICPerMed	International Consortium for Personalised Medicine
ICF	International Classification of Functioning, Disability and Health
LPGA	Loi fédérale sur la partie générale du droit des assurances sociales
NRP	National Research Program
SAHS	Swiss academy of humanities and social sciences
SAMS	Swiss academy of medical sciences
SCHEI	Swiss Conference of Higher Education Institutions
SERI	State Secretariat for Education, Research and Innovation
SNSF	Swiss National Science Foundation
SSC	Swiss Science Council (1965–1999 and again since 2018) 2000–2013: Swiss Science and Technology Council (SSTC) 2014–2017: Swiss Science and Innovation Council (SSIC)
SPHN	Swiss Personalized Health Network
SWR	Schweizerischer Wissenschaftsrat (1965–1999 und erneut seit 2018) 2000–2013: Schweizerischer Wissenschafts- und Technologierat (SWTR) 2014–2017: Schweizerischer Wissenschafts- und Innovationsrat (SWIR)
UK	United Kingdom
USA	United States of America
WHO	World Health Organization

Annex:

Health and disease in the era of big data

Concept analysis by Prof. Dominic Murphy,
University of Sydney, on behalf of the SSC

Health and disease in the era of big data

About the author

Dominic Murphy has been a professor of philosophy at the University of Sydney since 2012. He heads the Sydney Centre for the Foundations of Science[137]. His research, currently funded by the Australia Research Council, focuses on the philosophy of the cognitive and biological sciences. He has published over fifty articles and chapters in high-ranking journals and books on the history and philosophy of biology and medicine, bioethics and moral psychology. His current research interests are directed at the topic of self-knowledge and introspection.

Dominic Murphy studied philosophy in Dublin and London and earned a doctorate in philosophy at Rutgers University in 1999. After two years of postdoctoral fellowship in the Philosophy-Neuroscience-Psychology Program of Washington University in St. Louis, he became an assistant professor at the California Institute of Technology in 2000. In 2008, he joined the University of Sydney, where he was promoted to associate professor and director of the Unit for History and Philosophy of Science in 2012.

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137   https://sydney.edu.au/arts/our-research/centres-institutes-and-groups/
       sydney-centre-for-the-foundations-of-science.html.

Executive summary

Aim
The present study outlines the current state of the scientific debate on the various conceptions of health and disease from a philosophical point of view and discusses the importance of data-centric scientific approaches for conceptual questions.

Disease
Overall, much more research has been devoted to understanding disease than health, and accounts of disease have been concerned to a higher degree with trying to capture a scientific or clinical concept. Most philosophical positions recognise that both empirical and normative judgements enter into the concept of disease. Different conceptualisations can be characterised as either "naturalist" or "constructivist" depending on the relative priority given to empirical versus normative elements. Another criterion for distinguishing between "conservative" and "revisionist" disease conceptions is the extent to which common sense and scientific concepts may contradict each other.

Conservatism versus revisionism
Science might discover that people currently seen as healthy are in fact suffering from a physiological malfunction. A conservative position would recommend refining the analysis to ensure that the concept of disease does not cover this case, thereby maintaining a level of agreement between common sense and the medical consensus. A revisionist position would estimate that the case falls under the concept of disease, even if such a judgement appears counterintuitive to everyday experience.

Naturalism versus constructivism
Naturalists have theories about the functioning of a "human animal" and consider departures from that functioning the origin of disease. Such processes typically cause harm or impede normal life, but this is not the primary reason for recognising them as pathological. In contrast, in the constructivist view, we first circumscribe and identify a condition as deviating from the norm, and therefore we start looking for a biological cause for the disease. In this sense, a constructivist might still acknowledge the possibility of objective biological knowledge of the processes that cause the problems that we call disease. Constructivists are especially interested in tracing how social factors shape and modify categories over time. They also frame disability to a large degree as a social issue and see difficulties accompanying this condition as the result of the way the world is arranged. Overall, a constructivist's main problem is specifying the nature of the class of norm-violations that are medically relevant, while a naturalist's main problem is specifying the nature of malfunction.

Function
As the dominant model underlying contemporary medicine, naturalism assumes that it is possible to discover the correct functional composition of human physiology. However, functional models are viewed as problematic, as many present-day scholars believe that goal-oriented explanations introduce normative considerations into the foundations of medicine. Therefore, modern scholars turn to the concept of mechanism as a causal, rather than a goal-oriented concept of function. A mechanistic understanding of function requires that structures can be identified and analysed in terms of their contribution to the overall maintenance of the organism as a living system. Malfunction can then be conceived as a failure of the system to function as it is naturally supposed to. This raises several questions: how to be sure that a putative malfunction is not in fact contributing to a different property of the system? How to draw the boundaries of a mechanism and how to delineate what counts as the system? Some authors consider the biomedical concept of normality as an idealisation designed to let us classify real systems according to their departure from the ideal. Another issue is that for many conditions, variation between the normal and the pathological is continuous. In such cases, any partition of human conditions into classes involves some arbitrary decision.

Health
A simple view may be that health is the normal functioning of the human body and that a healthy individual is just someone whose biology works as our theories say it should. Most scholars, however, would add some components usually related to the aspect of quality of life. The phenomenological approach tries to capture the experience of being healthy. Other approaches consider the healthy person as being in harmony with his or her natural and social environment or apprehend health as adaptability, energy or sense of coherence. The instrumentalist conception points to the value of health as a way to achieve one's goals, either defined in terms of needs or in terms of ambitions and desires. Not everybody, however, equates health with well-being; rather, health is seen as a component of well-being. A concrete issue linked to the conception of health as well-being is the challenge of distinguishing between therapeutic and "enhancing" medical interventions.

Culture and gender

How health and disease are expressed may be affected by people's understanding of themselves and their place in the world. Cultural factors may influence the responses to pathogenic stimuli. Such mechanisms should be seen as different from the biological adaptation of human lineages to given historical conditions, such as the availability of animal milk as a food source in some human populations. Similarly, in order to comprehend the role that gender plays for health, we must discriminate between variances tied to female and male physiology on the one side, and the medical consequences of women and men occupying different social positions over the course of history on the other side. As a general rule, one has to be aware that most scientific explanations assume a representation of an exemplary human being that happens to be a man of European descent.

Theory

The survey concludes that there is no overarching theory of health and disease, in the same way, for instance, as the evolution theory explains how the variety of living organisms came to exist on earth. Such a framework would be most useful as a way of organising health data and drawing boundaries between biological systems. The life history theory, according to which an organism is designed to optimise survival and reproduction, could be tested as a framework for providing a context and purpose to the ever-expanding data. Comparable to the prevalent biostatistical model, the life history theory identifies conditions such as senescence as medically relevant, even though they might be normal in older cohorts. Nevertheless, such a plainly revisionist approach could accelerate medicalisation by creating new disease categories challenging common sense.

1
Introduction

It is unclear when the era of big data began. Beniger (1986) argues that the introduction of railways in the 1830s led to the rise of modern information technologies, which then grew apace after 1880 with full industrialisation. Machine industry involves huge, fast flows of goods, so it requires extensive information technology to be effectively managed. Beniger ignores the rise of mass armies, themselves moved by rail, in the 1860s (Howes 1998) but emphasises two great "discontinuities" (p. 23), involving first the railways of the 1830s and then an increase in bureaucratisation and control in the 1870s and 1880s. The first part of the economy to move at industrial speed were the railroads, and the accompanying jump in the size of the information sector is dramatic. So industrial production required improved information technology and improved information technology made new industrial developments possible. These developments undoubtedly generated large amounts of data with a corresponding increase in the segment of the labour force devoted to managing information. However, it seems uncontroversial to state that very recent developments have produced an increasing flood of specifically scientific data that is taken to have potentially "game-changing" (Leonelli 2017) consequences for the sciences, perhaps especially biomedical science. Although we may expect a great many changes in scientific practice as a result of these developments, their conceptual significance is hard to resolve. This report does not discuss research practice but attends to the concepts of health and disease as they underpin biomedical research. Much of the report is taken up with an overview of the existing philosophical literature to identify the conceptual issues that confront us. I suggest that few of these conceptual issues could be directly resolved by big data. However, I end by suggesting that there are important questions about ways in which new practices, enabled by big data, might be experienced socially. I do not think there will be new conceptual options that are due to the data revolution, but some existing possibilities might become more attractive.

2
Concepts of disease

Kitcher (1997, pp. 208–209) helpfully lays out the conceptual terrain:

"Some scholars, naturalists about disease, think that there are facts about the human body on which the notion of disease is founded, and that those with a clear grasp of those facts would have no trouble drawing lines, even in the challenging cases. Their opponents, constructivists about disease, maintain that this is an illusion, that the disputed cases reveal how the values of different social groups conflict, rather than exposing any ignorance of facts, and that agreement is sometimes even produced because of universal acceptance of a system of values."

Kitcher's objectivism is more often called naturalism in the recent literature. I will follow that usage. I begin with Kitcher's claim that a naturalist analysis is "grounded" on facts about the human body, as a way to distinguish the main approaches. Note that I am assuming that we are searching for a concept that will unify our thinking about medical disease. One could, though, be a pluralist, and assume that different concepts apply in different contexts. However, I do not think that the issues raised below would be affected by this. The big problem for naturalism is specifying the nature of malfunction, and the big problem for constructivism is specifying the nature of the class of norm-violations that are medically relevant. There is a way in which the two approaches could co-exist, though, that I will revisit at the end.

2.1
Naturalism and constructivism

The consensus among recent theorists has been that to attribute a disease to a patient involves making two judgements. These comprise 1) empirical judgements about human physiology (or, in the case of mental illness, psychology) and 2) normative judgements about behaviour or well-being (Bloomfield 2001, Boorse 1975, Culver and Gert 1982, Thagard 1999). The dispute between naturalists and constructivists can be interpreted as a dispute over 1) whether either judgement is fully objective and 2) which judgement has conceptual priority. The naturalist position is that diseases are destructive processes in bodily organs which "divert part of the substance of the individual from the actions which are natural to the species to another kind of action" (Snow 1853, p. 155; for discussion see Whitbeck 1977, Carter 2003, Broome 2006). The normative judgement is subordinated to the medical one; we recognise that pathological processes are of human concern because they typically cause distress or impede normal human life.

Now, this second type of judgement obviously involves normative criteria, to do with the extent to which a life is unnatural, undesirable or failing to flourish in some way. (There is not a clear consensus among writers here.) One important and controversial question is whether the judgements we make concerning our biology are also normative in some way. A further question concerns the relationship between the two types of judgements, in both medicine and common sense. Roughly, we can see the naturalist position as insisting on the priority of the scientific perspective on human nature; we have theories that tell us how a functioning human animal should be, and departures from that functioning are the basis of disease. This reflects a broader naturalism in philosophy that stresses its continuity with science. The constructivist position, on the other hand, thinks that our judgements about well-being and deviance form the basis of our attributions of disease: that disease is first and foremost a normative matter. Similar clashes over the normative occur in the context of disability. The "social model of disability" holds that disability is simply difference, and the difficulties and impediments that come with it are the result of the way the world is arranged. On this view, a wheelchair-bound person suffers from a physical environment without ramps or elevators, just as a black person might suffer from a racially hostile environment. But the lack of mobility should not be seen as an impairment or dysfunction any more than the black skin. Such views have particular policy consequences with respect to genetic testing or other pre-natal interventions. Some theorists worry about the possible eradication of difference via genetic manipulation, and others affirm the right of parents to produce children who resemble them, even if that means altering the foetus to (e.g.) induce deafness (Savulescu 2002, Parens and Asch 1999, Chadwick and Levitt 1998).

The social model of disability echoes an important strain in recent constructivist scholarship which suggests that our normative judgements alone determine who is sick or healthy. It is less widespread in philosophy than in other areas of the humanities and social sciences (e.g. Kennedy 1983, Brown 1990). This approach sometimes ignores any distinctively biomedical viewpoint and just examines norms. However, one might think there could be a set of naturally occurring processes, fully amenable to objective scientific study, that are held together in virtue of our interest in grouping them as a class. Kinds that work like this include "weed", "precious metal" or "vermin" (Murphy 2006). The labels indicate a class of phenomena that are unified in virtue of distinctive human interests. The properties of the members of such a class are not just subjective or normative. They can have entirely objective natures discoverable by science and manipulated through technology. But what makes them objects of manipulation is the significance they carry for human projects. The existence of the superordinate class depends on human interests but the subordinate members are natural kinds whose natures can be investigated scientifically. Adopting a perspective like this would let us see constructivism about disease as driven by normative concerns but still able to make room for scientific inquiry that promises real benefits. This view is not normally adopted by constructivists, who tend to deny that these processes can be identified independently of human values by, for example, the life sciences. Constructivist conceptions of disease are typically normative through and through, although the precise account of the norms varies between scholars.

I suggest that we should see two different dimensions; one concerning the relative priority of judgements of deviance/distress versus judgements of malfunction, and another concerning the scientific status of judgements of malfunction. If disease concepts are like "weed" then one can combine a norm-driven account of disease with an impeccably scientific account of the biological basis of disease. (There is also a third dimension, between conservative and revisionist positions, that I shall mention shortly.)

The crucial difference between the positions then is that, for a naturalist, diseases are objectively malfunctioning biological processes that cause harms. For a constructivist, diseases are harms that we blame on some biological process because it causes the harm, not because it is objectively dysfunctional. However, I have argued that a constructivist could acknowledge the possibility of objective biological knowledge of the processes that cause the problems that we call disease.

The crucial difference then is the relative priority, for our concepts of disease, of the scientific and the normative. I will look at each approach in turn, after introducing the other distinction I wish to insist on.

2.2
Revisionism and conservatism

On balance modern medicine looks naturalist. One question, then, concerns the extent to which common sense and biomedical concepts are related. There is little reason to expect scientific and common-sense concepts to agree in general, so if medicine and everyday thought disagree about disease, we may ask which concept should be adopted.

A revisionist naturalist argues that we should follow the science where it takes us and come up with concepts that further scientific inquiry even if that means that we eventually use the language in ways that look bizarre from our current standpoint of current common sense. But a revisionist could argue that our concepts should be reformed in the service of other, non-epistemic goals, such as emancipation for hitherto oppressed groups.

Naturalists tend towards conceptual conservatism. They typically appeal to our intuitions about illness as support for their own emphasis on underlying bodily malfunction. This assumes that our current concept is in good shape and that common sense and medicine share a concept of disease, and that medicine should respect ordinary judgements about what is and what is not a disease. Like many philosophers who think about other concepts that have both scientific and common sense uses, conservative naturalists about disease accept that folk concepts specify what counts as health and disease.

Revisionists argue that facts about physiological and psychological functioning, like other biological facts, can be obtained independently of human conceptions of the world. Science might discover that people we currently think are healthy are really suffering from a biological malfunction. In that case, a conservative would recommend finessing the analysis to ensure that the concept of disease does not cover this case. A revisionist could say that we must bite the bullet and judge that this case falls under the concept even if that judgement is counterintuitive. A revisionist naturalist regards health and disease as features of the world to be discovered by biomedical investigation, and therefore loosely constrained, at best, by our everyday thinking about health and disease.

2.2.1
Constructivism

Constructivism is hard to define satisfactorily, for two reasons. First, its core claim is a denial of the naturalist thesis that disease necessarily involves bodily malfunction. There are many views one might hold about the nature of the biological processes involved in disease, and many ways to deny it, so the positive constructivist claim varies across theories and is often elusive.

The second reason why constructivism can be an elusive target is that it has often stressed the role that value judgements have played in medical practice, or on the prevalence of culturally specific disagreements. Constructivists often seek to reconstruct the concept of disease as revealed by our practices. This stress on our practices is a common constructivist trope, whereas naturalists more often seek to analyse a concept so as to clarify what disease really is, however fumbling and biased our attempts to uncover it. Constructivists are often interested in tracing the social processes by which categories are formulated and changed over time. Conrad (2007, p. 7–8), for example, says he is "not interested in adjudicating whether any particular problem is really a medical problem… I am interested in the social underpinnings of this expansion of medical jurisdiction". But constructivists often present their theories as unmasking common sense or medical conceptions of disease, and hence as a kind of revisionism. They may accept that diagnoses of ill-health involve objective facts that people appeal to, or presume that they can appeal to, when they say that somebody is sick. Constructivism could, however, be a conservative view, aimed at uncovering our folk theory of disease as a pattern of behaviour or bodily activity that violates social norms.

The key constructivist idea is that concepts of health and disease medicalise behaviour that breaks norms or fails in some way to accord with our values; we don't like pain, so painful states may count as diseases: we don't like fat people or drunks, so obesity and alcoholism count as diseases. The assumption in a society might be that germs or other medically relevant causal factors are present in a person and have given rise to visible phenomena that indicate ill-health. But the constructivist says that our search for the relevant biological findings is undertaken because we have already decided that very overweight people are physically unappealing and we are trying to find a set of medically significant properties in order to make our wish to stigmatise them look like a medical decision rather than a moral or aesthetic one. The crucial constructivist claim is that we look for the biological facts that ground disease judgements selectively, based on prior condemnations of some people and not others.

Problems for constructivism

Constructivism looks correct as a historical point about some diagnoses that we do not accept any more; that is, we have thought that some human activities or traits were pathological because of values rather than scientific evidence – an obvious example is homosexuality which was only removed as a psychiatric diagnosis in the West recently and persists in some other countries. The diagnosis of greensickness was widespread in medieval and early modern Europe (King 2004); its symptoms included physical weakness, digestive irregularities, lack of menstruation and, allegedly, a change in skin colour. Because it was believed to largely afflict unmarried women, and to be cured by (legitimate, that is, marital) sex, it made virginity and puberty into medical problems. There may have been genuine medical problems in these cases, but the diagnosis expresses a discredited way of thinking about women, rather than a medical discovery. So constructivism is right as a historical report of some past medical practices. However, it is another task to show that constructivism is correct about our concept of disorder.

The chief problem for constructivism is that we routinely make a distinction between the sick and the deviant, or between pathological conditions and those that we just disapprove of. Our disease concepts cannot just be a matter of disvaluing certain people or their properties. It must involve a reason for disvaluing them in a medical way rather than some other way. Illness has never been the only way to be deviant.

We routinely judge that people are worse off without thinking they are ill in any way – for example, the ugly, the poor, or those in self-destructive relationships. We don't treat these judgements as a prelude to medical inquiry, so why do we do so in some other cases? Physically or mentally ill people, even if they are norm-breakers, are a distinctive class of norm-breakers. What's distinctive about them?

Notice that the problem is not just one of establishing that someone is badly off or is in some other disvalued state. Rather, the trouble is caused by the requirement that someone is badly

off in a specific, health-related or medically significant way. Rachel Cooper, for example (2002, pp. 272–274), analyses the concept of disease as a bad thing to have that is judged to require medical attention. She deals with the objection that specifying when someone is badly off is very difficult. Cooper admits that it is a hard problem, but replies that it is a widespread problem, which crops up in many areas of moral philosophy. That's true but it does not help to isolate the specific class of ways of being badly off that we regard as medically relevant.

The naturalist has a proposal viz. that conditions which doctors treat are diseases in so far as they involve natural malfunctions. The constructivist view is that the class of what we call malfunctions is picked out by its involvement in medical practice, and not the other way round. But they are unable to distinguish medical practices from non-medical ones.

The constructivist needs to explain why the value judgements that we direct at putative sick people form a special class of value judgements. And that explanation has to show, in addition, why we think the conditions that we single out as diseases with those special value judgements are candidates for a particular set of causal explanations. It's all very well to point out, as Reznek does (p. 88) that an aetiology only explains a pathology if we have already decided that it is a pathology. Even if correct, this dodges the conceptual question of why we first decide that only some people or conditions are pathological.

This problem for constructivist accounts is not one that can be resolved by extra data. It might be possible to make better and better predictions about, or more finely resolved discriminations among, categories of norm-defying behaviour. But the data cannot tell us which among these problematic conditions are the medical ones.

The naturalist has an answer. She says that we think some people are worse off because of a special kind of causal process, namely a disturbance of normal physical or psychological processes. It is that causal judgement that has conceptual priority, even if, as a matter of timing, the violation of a norm is what is detected first.

2.3
Forms of naturalism

Suppose we have decided that someone's biological systems do not function properly. We need to decide how to think about that person's condition. Naturalists usually admit that there is more to the concept of disease than biological malfunction even if they think that biological malfunction is a necessary condition for disease.

Normative considerations, on this account, inform our judgements about disease but do not have the conceptual priority accorded them by constructivists. We make judgements that someone is suffering in ways we associate with inner malfunction. We also see people who are suffering but who we don't think are ill or injured, because we do not regard their bodily dysfunction or damage as symptoms of disease: grief, vaccination, surgical incisions or childbirth are examples.

The idea is that whether someone's body is not functioning correctly is a separate question from whether it is bad to be like that. The second stage, the question about whether life is worsened by a malfunction, is omitted by simple naturalists who say that all there is to disease is the failure of someone's physiology (or psychology) to work normally. The view has few adherents, but Szasz (1987) uses simple objectivism about disease to support his claims that mental disorder is a myth.

2.3.1
Specifying causes

A generic naturalism says judgements of illness are sensitive to causal antecedents of the right sort, as well as to value judgements about the effects of those causes. What are the right causal antecedents? This is the hard problem for naturalists, since we need a way of specifying the biological causes of disease in a way that does not make them subjective.

Boorse (1975, 1976, 1997) and his followers contend that disease necessarily involves biological malfunction. Boorse distinguished "disease" from "illness". The former is the failure to conform to the "species-typical design" of humans, and the latter is a matter of judgements that a disease is undesirable, entitles one to special treatment, or excuses bad behaviour. An account of malfunction must be parasitic on a theory of function. Boorse sees functions as contributions to survival and reproduction (1976, pp. 62–63). Disease is failure to function as designed. Boorse's position has been very influential.

But the Boorsian view also faces difficulties. Cooper (2002, p. 265) suggests that Boorse's straightforward appeal to dysfunction must be qualified in the light of some apparent counterexamples. Although a woman taking contraceptive pills, for example, may be interfering with her normal functioning, this is not a disease. Boorse, though, could call it a self-inflicted disease that does not make the woman ill. Cooper also raises the problem of individuals with chronic conditions that are controlled by drugs. But, as Cooper sees, the big problem faced by Boorsian accounts is that of coming up with an acceptable conception of function in the first place.

Functions

The Boorsian analysis is of a common-sense concept of disease which bottoms out in a notion of malfunction as the cause of illness. Boorse's view is that conceptual analysis determines the empirical commitments of our disease concepts. It then hands over to the biomedical sciences the problem of finding the pertinent biological functions and malfunctions. Some recent theorists, notably Wakefield, have argued for an evolutionary account of function in terms of a system's replication via natural selection. However, the stress on a distinctively evolutionary account of function is unattractive, since the biomedical sciences employ a different conception of function. Two other problems affect the whole naturalist community. A reliance on scientific, functional decomposition as the ultimate justification of judgements of health and disease requires a revisionist, rather than a conservative, account. Also, it may not always be possible to settle contested cases by an appeal to a notion of normal human nature, because that notion is itself contested.

First, suppose we consider that the relevant concept of function is an adaptive one, and that dysfunction is a failure of a biological system to fulfil its adaptive function. This analysis of function is often termed the etiological account, and although it is widespread in philosophy of biology it seems to be conceptually tied to evolutionary fitness (understood as reproductive success) rather than health (Méthot 2011). Advocates of a thoroughgoing evolutionary approach to medicine (such as

Gluckman, Beedle and Hanson 2009) can be read as providing a framework within which we can make sense of processes that affect health and disease, but not as offering analyses of health and disease that are tied to fitness. Gluckman et al. (p. 5) consider the difference between lactose intolerance, which develops after weaning and is normal for most human populations, and congenital hypolactasia, a condition in which new-borns cannot digest maternal milk. The former is a consequence of the absence of pastoralism in most historical human populations and does not affect fitness in those populations, whereas the latter would have been fatal in the past and thus was selected against. However, they do not define disease in terms of fitness-lowering: they note rather that an evolutionary perspective can make us sensitive to hitherto neglected causes of pathology and also sensitive to over-hasty judgements of pathology in cases where the condition is normal among populations with a given evolutionary trajectory.

Perhaps in some areas of biology functional ascription is indeed teleological. However, Schaffner (1993) has argued very convincingly that although medicine might use teleological talk in its attempts to develop a mechanistic picture of how humans work, the teleology is just heuristic. It can be completely dispensed with when the mechanistic explanation of a given organ or process is complete. Schaffner argues that as we learn more about the causal role a structure plays in the overall functioning of the organism, the need for teleological talk of any kind drops out and is superseded by the vocabulary of mechanistic explanation, and that evolutionary functional ascriptions are merely heuristic; they focus our attention on "entities that satisfy the secondary [i.e. mechanistic] sense of function and that it is important for us to know more about" (1993, p. 390).

Schaffner is arguing that the biomedical sciences employ a causal, rather than a teleological, concept of function. This is in the spirit of Cummins's (1975) analysis of function as the causal contribution a structure makes to the overall operation of the system that includes it. Cummins's concept of function is not a historical or evolutionary concept. According to Cummins, a component may have a function even it was not designed or selected for and, therefore, parts with no selection history can be ascribed a function.

Most theorists who have attended to biomedical contexts agree with Schaffner that the function of an organ or structure can be understood without thinking of it as an adaptation. Medical understanding requires that functional structures can be understood in terms of their contribution to the overall maintenance of the organism as a living system. Explanation in medicine takes a model of the normal realisation of a biological process and uses the model to show how abnormalities stem from the failure of normal relations to apply between components of the model. This requires a non-historical function concept, one that is at home in causal-mechanistic, rather than evolutionary, explanation.

Mechanisms

In recent years philosophers have stressed the way in which explanation in the life sciences depends on finding mechanisms (Bechtel and Richardson 1993; Craver 2007; Tabery 2009). Rather than seeing explanation as a search for laws, mechanists seek the components within a system whose entities and activities are organised to produce the phenomena we want to explain. Philosophers disagree over exactly how to characterise mechanisms, but it is agreed that mechanisms comprise component parts and these parts have characteristic activities.

Perhaps the first and most enduring mechanistic explanation in the life sciences was Harvey's (1628) discussion of the pumping of the blood by the heart. The heart has anatomically distinguishable components: these include the left and right atria, the left and right ventricle and the valves between them. We might also include the veins and arteries, as they reach the heart, as part of the system, or we might not. If we were to consider the entire circulatory system we would certainly include the pulmonary vein and artery, the vena cava, the aorta and the capillaries. Bechtel (2006, p. 30) argues that the blood is also part of the mechanism, though a passive one. Explaining circulation involves identifying physical structures, such as chambers, valves and blood vessels and showing how they interact to cause the passage of the blood around the body.

The goal of finding out how a biological system works is fixed by our interests in health and well-being, but the naturalist assumption is that the goal is met by discovering empirical facts about human biology, not our own, culturally defined, norms. So, we diagnose someone as suffering from mesenteric adenitis not just because they are in discomfort due to fever, abdominal pain and diarrhoea, but because the lower right quadrant of the mesenteric lymphatic system displays abnormal inflammation. This thickening of the nodes is not just the cause of the discomfort, it is an objective failure of the lymphatic system to make its normal contribution to the overall system. For the naturalist programme to work, the biological roles of human organs must be natural facts just as empirically discoverable as the atomic weights of chemical elements. That may result in the overturning of common sense.

This raises a further issue. It is widely believed that function concepts are intrinsically normative, since they are teleological. Therefore, the objection continues, claims about natural function and malfunction introduce normative considerations into the foundations of medicine, which are supposed to be purely scientific on the naturalist account.

The view that the correct functional decomposition of humans can be discovered in nature is very strong. It contends that natural functional standards for human nature exist independently of what people think. Whereas the rules of kinship or gender are human conventions and vary across times and places, the way your heart or liver ought to work is a natural fact. The idea is that in cases where we can ascribe function to a physiological mechanism the standards of good performance are supplied by nature and not by human values. If that can be done, then malfunction can be understood as a failure of the system to function as it is naturally supposed to.

Whether or not this should be seen as normative, it is not the socially relative normativity appealed to by constructivists. The idea is that the norms already exist in nature, waiting to be discovered. The ways in which complex natural systems ought to behave depend on their role in the overall economy of nature – the olfactory bulb of the locust would have a job to do even if humans never existed. The crucial point is that in the life sciences, some biological system can fail to behave as a theory predicts without impugning the prediction: we can say that the system is malfunctioning. This contrasts with other sciences, in which, if a system fails to behave as predicted, the fault lies with the science, not the system. However, teleology is long out of fashion in biology, so an account of what makes the system malfunction will have to be supplied in a naturalistic form.

So where is an account of malfunction to be found? Supporters of a selectionist account of function advertise the ease with which an account of malfunction follows from the theory as one of its virtues. Their idea is that we can say when a system is malfunctioning by observing that it is not carrying out the job which natural selection designed it to perform.

In contrast, it is widely believed that it is harder to account for malfunction on systemic or mechanistic accounts of function. The argument goes that what a system is taken to do is relative to our explanatory interests. As Bechtel (2015) stresses, identifying the boundaries of the mechanism involves a strong element of idealisation. Biological mechanisms are as a rule causally connected with surrounding systems. The boundaries we draw reflect hypotheses about the best way to delineate natural phenomena for some explanatory purpose. What count as the boundaries of a mechanism relative to one explanation might be ignored in another explanation, in which the overall system gets partitioned very differently. So what counts as the system seems to be sensitive to our explanatory purposes, which introduces a clear element of human concern into a supposedly objective process. Identifying the mechanism seems subjective, which means that the identification of the malfunction seems subjective in the same way.

Godfrey-Smith (1993) argues that systemic concepts of function do permit attributions of malfunction. He argues that a token component in a system is malfunctioning when it cannot play the role that lets other tokens of the same type feature in the explanation of the larger system.

But this leaves unaddressed the issue of how we determine normal function. Wachbroit (1994) argues that when we say that an organ is normal, we employ a biomedical concept of normality that is an idealised description of a component of a

biological system in an unperturbed state that may never be attained in actual systems. Wachbroit argues that the role of normality in physiology is like the role that pure states or ideal entities play in physical theories. Statistically, a textbook heart, for example, may be very rare indeed. But it is the account of the organ that gets into the physiology textbook. The textbook tells you what a healthy organ is like by reference to an abstraction – an idealised organ. This concept of normality is not justified by appeal to a conceptual analysis that aims to capture intuitions about what's normal. It draws all its authority from its predictive and explanatory utility: against the background of assuming normal heart function, for example, we account for variation in actual hearts (a particular rhythm, say), by citing the textbook rhythmic pattern (which may be very unusual statistically) and identifying other patterns as arrhythmic. The point of textbook depictions of human physiology is to identify an ideal system that enables us to answer "what if things had been different" questions (Woodward 2003, Murphy 2006). The role of an idealisation, in this system, is to let us classify real systems according to their departure from the ideal. So normal human biological nature, in this sense, is an idealisation designed to let us impose order on variation.

Variation in biological traits is ubiquitous, and so establishing whether a mechanism is functioning normally is difficult: nonetheless, biologists seem to do it all the time. But not all diagnoses can be tied to a break between normal and abnormal functioning of an underlying mechanism, such as a failure of the kidneys to conserve electrolytes. Some conditions involve cutting between normal and pathological parts of a continuous variation, even in the absence of clear underlying malfunctions that separate the populations.

The more of this we do, the more we will have to complicate the analysis by appeal to risk factors and behavioural difficulties rather than natural standards of underlying function. This is a situation in which more data may help to resolve an issue. The challenge is to partition human beings into statistical classes that reflect levels of risk or physiological performance. The concern, though, is that these classes are only as legitimate as the judgements that go into their formulation. The concern is that the partition we produce will reflect contested conceptions of human flourishing rather than simple medical categories. Who gets to say what classes human beings should be divided into, for medical purposes?

Theory

As noted above, the attraction of the selectionist view of function is that it draws from a mature, well-confirmed scientific theory that can answer some of the queries I have noted. Evolution gives us a principled way of discriminating among functional and non-functional states and provides a (historical) basis for classifying biological phenomena. But it does not seem to

have the correct conceptual ties to health and diseases, since it is connected with fitness, not health or well-being.

One option in the face of these uncertainties is to adopt a fully revisionist account. Griffiths and Matthewson (2016) for example, defend a "selected effects" view according to which medically significant malfunction is just the failure of a system to function as it was selected to by evolution. However, their understanding of the context of evolutionary accounts of function is notably revisionist. They root their understanding of evolution in life-history theory, according to which an organism is designed by evolution to solve the problem of finding "the optimal way to parcel the resources available to it into offspring. This problem is modelled as the simultaneous optimization of two parameters, the probability of surviving to each age class and the number of offspring produced in each age class, integrated across all age classes (there are both continuous and discrete versions of these models)."

On this perspective, Griffiths and Matthewson offer a principled scientific reason for preferring an account of function and malfunction, but also give us a way of encompassing many conditions as objects of medical intervention even if they are not diseases in our customary understandings. Take senescence, for example. Many conditions, such as osteoarthritis, are statistically normal in older cohorts. Should they be seen as diseases in these groups? The life-history perspective gives us a way to understand this as a normal effect of aging in an organism which trades off greater fitness when young in return for less physical capacity in old age. But the life-history perspective also gives us a means of distinguishing different sorts of biological processes as "genuinely senescent phenotypes and age-specific adaptations to the ecological demands of later age classes".

A view like this allows us to make distinctions among reference classes based on evolutionary and environmental contingencies. In effect, it provides principled grounds for distinguishing ways of going wrong for an organism relative to our ordinary judgements. Some may be simple malfunction, others may be adaptive on a life-history perspective, while others may represent evolutionary lineages that are placed in an environment they are not well adapted for. Lactose intolerance, for instance, may not be a disease, but could nonetheless be a medical problem for members of non-pastoralist cultures (e.g. East Asians) who are trying to make a life in cultures where such adaptation is normal (as in many Western European environments). It may not be a disease, but Griffiths and Matthewson can still call it a medical problem and have good scientific grounds for doing so.

The life-history perspective, although billed as an account of disease, is not one. Rather, it is the basis for a scientifically motivated classification of grounds for medical intervention. In the final sections of the paper I will suggest that such an account may be especially well suited for the era of big data, but that such strong revisionism also brings problems of legitimation in the public sphere. Before I get to that, though, let me briefly look at health.

3
Health

3.1
Health and biology

Health is less studied than disease. Conceptions of health, like conceptions of disease, tend to go beyond the simple condition that one is in some biologically significant state. In the case of health, one view is that a healthy individual is just someone whose biology works as our theories say it should. This is the counterpart, in theories of health, to simple objectivism about disease. As with disease, however, most scholars who write about health add further conditions having to do with quality of life. On this view, we need a threefold distinction between disease, normality and health, where health involves some properties of a person's life that enable us to evaluate how well it is going for them. Carel (2007), for example, thinks that the important thing about health is one's lived experience of one's own body, and in particular, that one should not feel estranged or alienated from one's body. Carel argues that health should be understood phenomenologically as the experience of being at home in one's lived body, rather than merely the normal functioning of the body seen as a biological unit.

From the naturalist perspective, one problem with this proposal is that it ignores that one can feel perfectly at ease with one's lived body even if one harbours, unaware, a diseased system. Indeed, Carel argues that someone who is ill can be, in her sense, healthy, if they are adapted to their bodily predicament. From her perspective, objections like the one just mentioned miss the point, since they privilege a biological perspective rather than a phenomenological one. Her project is avowedly revisionist: she wishes to replace existing concepts of health with views that aim to capture the experience of being healthy (or unwell). It may well be that perspectives like Carel's are neglected in contemporary medicine, and especially important in disability studies. But it does not follow that the concepts of health and disease, rather than aspects of our practices that employ those concepts, should be reformed along the lines she suggests.

Carel's stress on experience is directly challenged by views like Gadamer's. He insists (1996, p. 113) that it is absurd to ask someone if they feel healthy, since health is "not a condition that one introspectively feels in oneself. Rather, it is a condition of being involved, of being in the world, of being together with one's fellow human beings, of active and rewarding engagement in one's everyday tasks." Gadamer's healthy person is someone who is in harmony with the social and natural environment, and disease is a disturbance of this harmony. Canguilhem (1991, 2012) thinks of health as flexibility, in the sense that a healthy organism can tolerate environmental impacts,

adapts to new situations and possesses a store of energy and audacity. This is not something that can be measured by physiology (2012, p. 49). Canguilhem's approach suggests what is wrong with Gadamer's objection to phenomenological accounts of health. There does not have to be a special feeling that is the feeling of being healthy. Rather, for a view like Canguilhem or Carel's, healthy people experience the world as an arena to express themselves in rather than a bunch of threats. A similar perspective, though closer to science, is perhaps Antonovosky's idea of salutogenesis. Antonovosky (1987) sees a sense of coherence as enabling one to understand the inner and outer environments and utilise physical and psychological resources to deal with stressors.

3.2
Instrumentalism

Gadamer's view is reminiscent of what Richman (2003) calls "embedded instrumentalist" theories, which claim that the value of health is that it is a way to achieve your goals: how healthy you are depends on how well you get what you want or need. Embedded instrumentalism is very popular. Nordenfelt (1995) considered two versions of this approach. One version defines the goals relevant to health as needs, which are understood as having a biological basis. Another view defines goals in terms of the ambitions and desires of the individual. Nordenfelt (1995, p. 90) argues that a healthy person is one who can satisfy her "vital goals", which are those that are necessary and sufficient for her to be minimally happy.

Embedded instrumentalist theories of health have an obvious appeal. Once we argue that health involves judgements about how well a person's life is going, we need a way to evaluate that, and an immediately attractive idea is that someone's life goes well if they can achieve their ambitions or satisfy their goals. An apparent difficulty, however, is that much the same terrain is covered by theories of well-being, and while people think that being healthy is important to their well-being (Eid and Larsen 2007), they do not identify the two. Rather, they think of health as a component of well-being.

Some embedded instrumentalist theories, though, appear to be in danger of defining health in such a way that it is synonymous with well-being. Richman (2003, pp. 56–57) supposes that someone is healthy if she can strive for a consistent set of goals that would be chosen by an idealised version of herself if she were fully aware of her "objectified subjective interest" (p. 45). That is, they are the goals she would choose if she had complete knowledge of herself and her environment and perfect rationality.

In this case it seems that a theory of health is in danger of becoming a general theory of well-being, and Richman does not discuss the relation between the two. A further complication is the relationship between medical interventions designed to cure diseases, and other medical interventions which are

"enhancement technologies" (Elliott 2003). The line between enhancement and therapy is very hard to draw: Harris (2007, p. 21) for example, uses the example of vaccination, which is both a therapeutic protection against infection and an enhancement of our natural immune system. Whitbeck (1981) defines health in terms of the psychophysiological capacities of an individual that support her "goals, projects and aspirations in a wide variety of situations". This suggests a view in which we can see biological capacities as at the core of health in so far as they help people's lives to go better. There seems to be a general sense among scholars that the important aspect of health is that it enables one to live in the world; to act unimpeded in the pursuit of one's projects.

3.3
Culture and gender

Malgri afflicts Aboriginal Australians who live on the Wellesley Islands in the Gulf of Carpentaria. Its typical symptoms include intestinal pain, bloating, vomiting and headaches. According to the local explanation, it is caused by eating terrestrial food and then going into seawater without washing in freshwater, or eating seafood and then going ashore without washing in salt water. The local spirit totem enters your body and makes you sick, but only if it does not recognise you. The coastline is divided into zones, each with its own totem animal. This explanation is not one that would satisfy medical criteria, but the symptoms are real.

A condition such as Malgri illustrates the way in which specific cultural forms of life may affect the expression of illness. It is not likely that Wellesley islanders share some unique biology that makes them suffer a diagnosis that no other human group endures. However, their understanding of themselves and their place in nature may affect the ways in which they express responses to pathogenic stimuli. We have already noted conditions like lactose intolerance, that do depend on specific evolutionary pathways that differentially influence some human lineages. We need both an awareness of such historic differences among human groups and an awareness of the specific cultural understandings that affect the expression of illness. Similar remarks apply to gender; there exist conditions that are tied to the physiology needed to produce eggs rather than sperm, but there are also medical consequences of women's subordinate social position that are not directly biological but are tied to health outcomes all the same.

The great danger, with both culture and gender, comes from the need for idealisation. I mentioned above that expla-

nation in biomedicine routinely involves idealisation. If one expects to explain an ideal or exemplary human being, it is important to be aware of the risks of taking one particular type of human (historically, men of European descent) as the typical case that should be explained, and either treating other types of human as pathological or attempting to explain them using illicit assumptions imported from what is taken to be the typical case. We should also (as Leonelli notes) be alert to the possibility that data gathering will neglect marginalised groups.

4
Interim conclusions

I suggest that data, however big, is unlikely to resolve any conceptual problems. There are clearly medical technologies (such as evidence-based) approaches for which data is central and where further data, properly managed, might indeed be a game-changer. However, I have argued that the array of existing concepts of health and disease face conceptual problems that are likely to remain even if torrents of new data come in.

In discussing Griffiths and Matthewson, though, I did stress the importance of theory as a way of organising data. We can make discriminations among the myriad forms of living things, for example, because we have a well worked-out theory of life on earth which explains why that variety of living things came to be. I think it fair to say that the survey of the conceptual terrain in biomedicine suggests that there is no overarching theory that underpins our thoughts about health and disease. There are local theories addressing problems in physiology, infection, development etc., but no overall scientific perspective that enables us to answer the questions about objectivity and normativity that I have pointed to.

One question, then, is whether things might look different if we had a theory that could play this overall, interpretive role of resolving the conceptual questions. Such a theory provides a context and purpose for the data and allow us to be clearer about what questions the data were being applied to. With a torrent of data there comes a large array of options for dividing the population into classes, including the ultimate subdivision of the population, for some purposes, into individuals, as imagined by personalised medicine. Some statistical divisions may have a technical or applied basis; we might discriminate between fast metabolisers of a drug and slow metabolisers, with a view to calibrating dosage. Other divisions will make sense against a theoretical background that explains why the data should be partitioned in one way rather than another.

5
Looking ahead

Suppose we imagine a life-history theory along the lines of that offered by Griffiths and Matthewson as a basis for establishing biomedical inquiry going forward. I choose this for its intrinsic interest, but also for illustrative purposes; I do not mean to suggest it is the only possible option. I have noted that it is fully revisionist, in the sense that it aims to taxonomise possible medical interventions rather than capture existing concepts of disease. The question such approaches raise is partly scientific and partly political.

A theory that is oriented to existing science as a principled way to use the data will be a successor theory to the one we have now, and like all successor theories it will involve a conceptual overhaul that makes some of our old projects and questions simply impossible to carry on. An account of health and disease which is answerable only to scientific criteria risks becoming alienated from the populations it is designed to serve, as it re-interprets forms of human suffering to fit bodies of thought that are not familiar within the culture. We may not recognise the new disease categories or understand the theory that motivates them. In some areas of medicine, this may make no difference, but in others it may intrude on important ways in which we currently understand ourselves. The truth about fermentation might be hard to grasp, but it does not interfere with your ability to enjoy a glass of wine. But we all care about our health and that of our loved ones, and it may not help us to be told about them in scientific language that is incomprehensible to us because it departs from existing categories that are familiar from ordinary thought. Medicine is obviously a scientific enterprise which requires a technical vocabulary, but it must also be communicable to the public. I think this is attractive as a moral matter – since people should know the truth about themselves – but also medically desirable to give people incentives to take care of themselves.

This is potentially a problem if we develop bottom-up models of disease that emerge from the generation of new data via new techniques. With new theoretical tools we might be able to adopt well-founded statistical partitions between human groups, including different diagnostic categories, based on genetic or other variation. But if the theory enables new data to reinterpret human beings too substantially, we will risk losing our grip on what matters to people. It is important to consider how we can make new developments accessible to the patients who stand to be affected by them. This might be a special problem for historically marginalised groups who have existing reasons to be suspicious of established medical structures. But we need to think very hard about how revolutions in medicine might be experienced by the people they are designed to serve. One thing the constructivist tradition got right was its stress on the moralised nature of disease categories.

Physical states can be healthy or not. Unlike mental states, they cannot be warranted or justified, but they can be understood as evidence of the moral status of the individual (cf. Arpaly 2005). Many diseases are understood as saying something about one's nature or moral worth (sexually transmitted diseases, for instance, or lifestyle diseases). A physician may think of a disease as a purely unhealthy or undesirable state, but it may be experienced by a patient as a moral judgement. If we are to develop new categories of disease we may find ourselves developing new forms of moralism without meaning to.

I am, in sum, sceptical about the likelihood of our conceptual difficulties being overcome by more data. However, it is possible to imagine that increased data could accelerate a trend towards greater medicalisation, as more and more information becomes available about human beings, their problems and the possible mitigation of those problems. With greater capacity to measure and differentiate human problems, the line may blur between disease and non-pathological but unwelcome states of affairs. We may see greater pressure to change people via personalised drug delivery or other treatments: greater ability to measure may lead to more desire for intervention on many conditions, regardless of whether those ways of being should be seen as diseases or not. If that does come about, the conceptual issues I have discussed here will be less pressing, and be replaced as urgent matters by moral questions about the aim of intervention and the best life for human beings.

6
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