SUMMARY PAPER

Review of EU initiatives related to the implementation of whole genome sequencing into clinical practice

European Union • 1+Million Genomes • Whole genome sequencing

Problem

Whole-genome sequencing (WGS) shows promise for stratifying disease management, identifying patients with rare disorders, improving prevention and identifying the causes and course of infections. This could improve both health and non-health outcomes for patients/citizens, thanks to more effective use of new technologies and (agnostic) medicines.

Currently, there is still a lack of evidence for the direct and indirect clinical benefits of WGS. As genomic testing is regulated at national levels, each country in the EU has different financial and infrastructural systems to implement WGS. This could lead to different diagnostic and treatment strategies, and therefore different (unequal) health outcomes across the EU. Health Technology Assessment (HTA) could support implementation by providing information regarding, e.g. cost-effectiveness, budget impact, organisational and patient-related issues.

Activities

- **Kick-off meeting** of WG6 HEOR (June 2019, Brussels). There were 9 participants with various backgrounds, such as Health Economics experts, rare diseases, common complex diseases, and oncology. In this first meeting, ongoing initiatives in genomics and HTA were explored from the different countries
- **Monthly virtual meetings** with of more than 20 experts from 18 countries. Several countries and industries presented ongoing initiatives and experiences (including France, UK, Spain, Netherlands, Finland as well as Roche and Illumina)
- **A survey** performed on sustainable finance models and outcomes underlined the immaturity of economic evaluations in genomics

Introduction

EU Member States' initiative '1+Million Genomes' aims to enable secure access to a cohort of at least 1 million sequenced genomes across the EU for healthcare, research and policymaking purposes. Healthcare implementation aspects of this endeavour are discussed within Working Group 6 'Health economics and outcome research' (WG6 HEOR).

Objective

To stimulate the development of a common approach to generate evidence for WGS application in clinical practice by providing an overview of ongoing European initiatives:

1. Specific clinical and public health indications and patient groups in which the implementation of WGS could be already valuable
2. Information on cost-effectiveness and budget impact of implementing WGS
3. Information on (management) practicalities (e.g. financing, organisational changes) of implementing WGS

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Lessons learned

According to the various initiatives in several countries, we explored similarities and differences:

Similarities – General interest in potential use for research purposes. Lack of standardised approaches (e.g. HTA), lack of consensus on quality measures, legal and financial solutions for WGS implementation, the need for genomic literacy improvement among all stakeholders (from patients, clinicians to society). Methodological and practical difficulties in developing cost-effectiveness studies, the need to move towards alternative forms of economic evaluation that also consider aspects other than health gains and costs, and the need to involve stakeholders in genomic medicine evaluations.

Differences – Evidence available and used: variability in evidence on the application of WGS in each healthcare system; population vs personal health approaches. Indications and extent of application for e.g. different clinical needs, different tumour types, use of different diagnostics in rare diseases. Practical and financial impact: decisions on infrastructure (centralised vs hospital-based), referral and pathways, and strategy of reimbursement (governmental, private funds, grants, etc.).

Added value

By identifying good practices, barriers, and solutions and providing guidance for implementation of WGS in clinical practice in EU countries, we should support national efforts to find the most optimal solutions and value for money implementation of WGS by:

- Contributing to standardised protocols, quality measures, legal solutions for genomic data storage, sharing and secondary usage
- Evaluating previous genetic medicine adoption models and the added value of gathering this evidence in several countries
- Helping support building evidence to inform policymakers in their decision for reimbursement and implementation of WGS
- Supporting further method development by stimulating and maintaining contacts with HTA-peers all over the EU
- Making inventories of main benefits for individuals and society

References and documentation

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