

Research Project

Ethical considerations in pediatric genome-wide sequencing

Project funded by own resources

Project title Ethical considerations in pediatric genome-wide sequencing

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Whole exome sequencing (WES) and whole genome sequencing (WGS) are increasingly used in health-care and research to identify genetic variations, assisting disease diagnosis and prognosis as well as treatment decisions. Besides many potential benefits of WES/WGS for medical care, it also involves a number of important regulatory and ethical challenges. However, there is currently a lack of empirical research on the ethical issues surrounding WGS/WES in Switzerland. It is crucial to investigate the country-specific attitudes, values, beliefs and resulting needs of the relevant stakeholders in Switzerland as an assessment of these is highly valuable for any policy guidance and ethical use of the techniques.

The study has the following main objectives:

A. Conduct empirical studies to inform and contextualise the ethical analysis;

1. Undertake a systematic qualitative literature review to gather evidence regarding the full spectrum of ethical issues involved in whole-genome/ whole exome-sequencing
2. Explore empirically the attitudes of clinical geneticists regarding WGS/WES in Switzerland and Germany
3. Explore empirically the attitudes of parents involved in WES of their children in Switzerland

B. Develop normative conclusions with regard to the regulation and practice regarding WGS/WES in Switzerland

The first objective is met by systematic qualitative review methodology (a) and qualitative research methodology (thematic analysis, b and c). For the latter two, semi-structured interviews in Switzerland and Germany are carried out with geneticists and parents of children involved in WES. These exploratory interviews are not based on a predefined understanding on how it should be dealt with WES/WGS of needs but seek to capture the needs from the perspective of clinical geneticists and parents involved.

For the second objective Empirical Bioethics methodologies will be employed. The normative analysis in this project will constantly accompany the empirical data collection. It will concentrate on the overall ethical research question of this PhD: How should be dealt with WGS/WES in an ethical manner in Switzerland – especially regarding children?

This thesis adds both to the growing international body of research on responsible and ethically justifiable uses and implementation of new technologies in genomics as well as to filling the gap of research on ethical issues regarding WGS/WES in Switzerland, thus providing key information for Swiss stakeholders in genetics and healthcare services.

Keywords bioethics, whole genome sequencing, whole genome sequencing, qualitative research, genetics, genomics, pediatrics, health politics

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