Next generation sequencing in diagnosis

Health Council of the Netherlands

Record Status
This is a bibliographic record of a published health technology assessment. No evaluation of the quality of this assessment has been made for the HTA database.

Citation

Authors' conclusions
The term "next generation sequencing" (NGS) covers a range of techniques that are designed to map a person's entire genetic code quickly and relatively cheaply. This horizon-scanning report explores the benefits and issues associated with the use of these techniques in diagnosis. There are various levels of sequencing. One level is represented by gene panels consisting of a limited number of genes associated with specific disorders, such as cardiomyopathies. At another level, all of the protein-coding sections of the DNA (the exons) are sequenced. This is referred to as whole exome sequencing (WES). WES is used in cases where a disorder (such as severe intellectual disability) is suspected of having a genetic cause, but where the defect involved has not yet been traced to a specific gene (or genes). Alternatively, the entire genome – including all coding and non-coding sections – can be sequenced. This is referred to as "whole genome sequencing" (WGS) and is still primarily used for scientific research. Both WES and WGS can be used to analyse just a part of the sequenced code, similar to a gene panel. Accordingly, sequencing and the analysis of sequences are not necessarily the same thing.

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Address for correspondence
Postbus 16052, 2500 BB Den Haag, The Netherlands. Tel: +31 70 340 7520; Fax: +31 70 340 7523 Email: info@gr.nl

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