

A scoping study to explore the cost-effectiveness of next-generation sequencing compared with traditional genetic testing for the diagnosis of learning disabilities in children

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Declared competing interests of authors: none

Published June 2015

DOI: 10.3310/hta19460

Plain English summary

Diagnosis of learning disabilities in children

Health Technology Assessment 2015; Vol. 19: No. 46

DOI: 10.3310/hta19460

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Next-generation sequencing (NGS) techniques are new developments in genetic testing that are expected to increase the number of health-related conditions that can be diagnosed. Currently, these techniques are mainly being used for research purposes. The purpose of this study was to find out whether these techniques offer value for money when used in the NHS to help diagnose learning disabilities in children. The study found that:

- NGS technologies are at an early stage of development and it is too soon to say whether they can offer value for money when used to help diagnose learning disabilities in children
- stakeholders had very different views about how, and over what time period, NGS technologies could, or even should, be used in the NHS
- if NGS technologies were to be introduced into the NHS:
 - more staff would be needed (particularly data analysts and genetic counsellors)
 - there would be a need for more genetics-related education for NHS staff and for members of the public
 - new systems would be needed to make sure that these new technologies were used safely and fairly.

ISSN 1366-5278 (Print)

ISSN 2046-4924 (Online)

Impact factor: 5.116

Health Technology Assessment is indexed in MEDLINE, CINAHL, EMBASE, The Cochrane Library and the ISI Science Citation Index.

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This report

The research reported in this issue of the journal was funded by the HTA programme as project number 12/47/01. The contractual start date was in April 2013. The draft report began editorial review in April 2014 and was accepted for publication in February 2015. The authors have been wholly responsible for all data collection, analysis and interpretation, and for writing up their work. The HTA editors and publisher have tried to ensure the accuracy of the authors' report and would like to thank the reviewers for their constructive comments on the draft document. However, they do not accept liability for damages or losses arising from material published in this report.

This report presents independent research funded by the National Institute for Health Research (NIHR). The views and opinions expressed by authors in this publication are those of the authors and do not necessarily reflect those of the NHS, the NIHR, NETSCC, the HTA programme or the Department of Health. If there are verbatim quotations included in this publication the views and opinions expressed by the interviewees are those of the interviewees and do not necessarily reflect those of the authors, those of the NHS, the NIHR, NETSCC, the HTA programme or the Department of Health.

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