Technoleg Iechyd Cymru
Health Technology Wales

Topic Exploration Report

This report summarises the existing evidence on the technology of interest in this Bevan exemplar application.

Topic:	**GENOTIME Project - genetic innovation for intellectual disability/developmental delay**
Topic exploration report number:	TER132

Aim of Search

Cedar researchers, on behalf of Health Technology Wales, searched for evidence on the use of DNA next generation sequencing (NGS) and array comparative genomic hybridisation (CGH) in children with intellectual disability (ID) or developmental delay (DD). The new technology combines the capability of these two tests into a single NGS test using an existing platform. The intention is to determine a genetic cause of DD/ID more quickly than currently available.

The adoption of this technology would result in a change in the patients' care pathway. Currently the array CGH test is ordered for patients by clinicians which are then referred to Clinical Genetics for the NGS test. The new single test would be ordered by clinicians without referral to Clinical Genetics.

Summary of Findings

No studies or clinical trials were identified that reported a single NGS test that combines array CGH and NGS in DD/ID. There are a range of genetic testing modalities; however, their relationship to the new technology is not clear.

Key Sources of Evidence

The Deciphering Developmental Disorders project is referred to by the topic proposer but the new test has not been identified (https://www.ddduk.org/).

Areas of Uncertainty

It is unclear whether more than one version of the proposed single NGS test exists, or whether this is a proprietary or generic technology.

Brief literature search results

Resource	Results
UK guidelines and guidance	
Healthcare Improvement Scotland	We did not identify any relevant information from this source.
NICE	NICE. (2017). Autism spectrum disorder in under 19s: recognition, referral and diagnosis [CG128]. Online: National Institute for Health and Care Excellence. Available at: https://www.nice.org.uk/guidance/cg128 [Accessed 13.08.2019]. Research recommendation: "What is the effectiveness and acceptability of comparative genomic hybridisation (CGH) array compared with current genetic testing in children and young people with identified autism?"
Guidelines International Network	We did not identify any relevant information from this source.
Secondary literature and economic evaluations	
ECRI	We did not identify any relevant information from this source.
Cochrane library	We did not identify any relevant information from this source.
Medline	We did not identify any relevant information from this source.
Primary studies	
Medline	We did not identify any relevant information from this source.
Cochrane library	We did not identify any relevant information from this source.
Ongoing secondary research	
Clinicaltrials.gov	NCT03857997 – Contribution of High Throughput RNA Sequencing Combined With Sequencing of Whole Genomes in the Diagnosis of Intellectual Disability (DI-WA) (France) Additional trials: • Deciphering Developmental Disorders (DDD) project - https://www.ddduk.org/ • Genomics England - https://www.genomicsengland.co.uk/portfolio/paediatrics-gecip-domain/

Date of search:	13 August 2019
Concepts used:	Array CGH, genomic, NGS, sequencing, intellectual disability, developmental delay