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Title Comparative genomic hybridization array (CGHA) analysis in postnatal context

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Aim

Comparative genomic hybridization array (CGHA) analysis is a molecular cytogenetic technology that is used to detect quantitative variations of the genome, corresponding to chromosomal material losses or gains (deletions, duplications, insertions, abnormal chromosome numbers, etc.) compared to a reference diploid genome. This pangenomic technology has a substantially higher resolution than standard karyotype, which is considered as the gold standard for whole genome analysis. A request was made for an assessment of this technique by HAS, from the French Ministry of Health and the National Health Insurance, with a view to permanent cover under common law.
CGHA was previously assessed in 2019 by HAS for use in cancer care.
The aim of this assessment was to determine the current benefit of CGHA use in the postnatal context, within the scope of routine care. This involved defining the indications of interest and the role of this technology in the diagnostic strategy, in the different clinical contexts in question, as well as the conditions of its implementation.

Conclusions and results

On the basis of the compiled and reviewed data, HAS has approved the reimbursement of this procedure:

- as a first-line in the diagnostic of: neurodevelopmental disorders (syndromic or nonsyndromic intellectual disability, syndromic or nonsyndromic learning disability, autism spectrum disorders, in particular when associated with intellectual disability, epilepsy, in particular when associated with a neurodevelopmental disorder, and polymalformative syndromes);
- as a second-line approach on a case-by-case basis: in the diagnosis of abnormal growth, isolated malformation and neonatal hypotonia; or to check a CNV detected using another technology, characterise chromosomal rearrangement identified by karyotype and test families to identify the hereditary or *de novo* nature of a rearrangement observed in a child.

Figure 5 contained in the conclusions (section 6) summarises the role of CGHA in the diagnostic strategy of the different clinical contexts discussed in the report.

Recommendations

This report recommends (section 6) a number of implementation conditions concerning the information required with the prescription, the procedure setting, the minimum CGHA resolution level, information to be contained in the report, contexts for which family testing is required for index case diagnosis, pooling of CGHA results in reference databases to facilitate interpretation, and information to be given to the patient.

Methods

The method used for this assessment was based on a critical analysis of the summarised literature (17 clinical practice guidelines, five health technology assessment reports and two systematic reviews) identified by a systematic search and selected on the basis of explicit criteria, collection of the collective opinion of relevant professional bodies, patient and user associations, as well as the collection of observations from public health institutions.

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