Is genetic evaluation of a child with either global developmental delay or mental retardation useful?

childhood disability LINK

Summary

Fluorescent In Situ Hybridization (FISH): A technique that can be used to detect and localize the presence or absence of specific DNA sequences using fluorescent probes that bind to only those parts of the chromosome with which they show a high degree of sequence similarity to the probe.

Molecular testing for Fragile X: Involves looking for an abnormal repeat in a particular gene on the X chromosome.

Metabolic studies: Involves looking for inherited metabolic disorders. Metabolic disorders are those that interfere with the body's ability to perform the many chemical reactions that are necessary for the maintenance of life (eg. the conversion of sugar into usable energy).

Cytogenetic studies: Involves primarily looking for structural changes in all the chromosomes (e.g. duplications, deletions and translocations).

Comparative genomic hybridization: A method for analyzing the deletions and duplications in a subject's DNA (e.g. often the DNA in cancerous tumors are analyzed using this method).

The main aim of this paper was to describe the best possible genetic testing that could be used for finding a cause for childhood developmental delay or intellectual disability. It was found that no single approach is consistently supported and a variety of valid testing options exist. In general, the literature does support looking at the family history over 3 generations. As well, a detailed neurologic examination aimed at determining abnormalities in head size and brain abnormalities is warranted, which could suggest the possible use of neuroimaging. The paper also found support for the use of the following genetic tests: cytogenetic studies (also known as high resolution karyotyping), fluorescent in situ hybridization (also known as FISH), molecular testing for Fragile X and particular metabolic studies in certain specific clinical situations. A future potential for the use of comparative genomic hybridization was also highlighted.

What families should know

Despite detailed assessments by specialists and subspecialists, a minority of children with developmental delay or mental retardation still remain without an underlying cause determined. More specialized genetic testing may play a role in the future in determining causes.

What practitioners should know

This paper provides an overview of what can be expected through the clinical genetic evaluation of a child with developmental delay or mental retardation.

Reference

Moeschler, J.B., Shevell, M.I. and the AAP Committee on Genetics (2006). Clinical genetic evaluation of the child with mental retardation or developmental delays. *Pediatrics*,117, 2304-2316.