Microarray scans of the whole genome give cost-effective diagnoses of global developmental delay

childhood disability LINK

Summary

Microarray analysis is a relatively new technology that gives a detailed picture of an individual's entire genome, highlighting anomalies too minute to be detected by traditional chromosome visualization techniques such as karyotyping. Existing guidelines for the diagnosis of global developmental delay (GDD) in children recommend microarray analysis as the first step, but, because of its high cost, many doctors continue to karyotype individuals for initial testing. Nonetheless, when microarray analysis is the first test performed, it can reduce the need for further diagnostic tests and increase the yield of diagnoses, making it more cost-effective overall.

This study retrospectively examined 114 children consecutively diagnosed with GDD at the Montreal Children's Hospital. For children who required genetic confirmation of the disorder, karyotyping was the first step, followed by subsequent tests if this gave a negative result. The cost of all tests performed for each child was collected and compared to the cost of performing aCGH alone. Tests that would be performed regardless of whether karyotype analysis or aCGH was performed first were excluded from cost calculations. A two-tailed student's t-test was used to compare the mean cost of diagnosis when karyotype was the first test to that when aCGH was the first test.

What families should know

GDD is characterized by severe delay in intellectual development, malformed features, or separate complications with at least two organ systems. It is most frequently caused by anomalies in chromosome number or structure. While chromosome number variations are easily detected by karyotype, structural variations in chromosomes may be too small to be detected this way. Microarray analysis has been shown to be much more effective for detecting genetic indicators of GDD, allowing diagnoses in 10-15% of children with clinical markers of GDD who show normal karyotypes.

What practitioners should know

GDD can often be diagnosed without genetic analysis by examining a child's medical history and current neurological and psychosocial function. However, when genetic evidence is required for a conclusive diagnosis, it is more time and cost-efficient to initiate genetic testing with array-based comparative genomic hybridization (aCGH), or microarray analysis, rather than with karyotype. Microarray analysis can give a complete and detailed picture of the genome, making other costly tests unnecessary. When genetic testing begins with microarray analysis, a diagnosis can be made within 10 days, allowing for earlier treatment of affected individuals. In contrast, testing that begins with karyotype may require further costly diagnostic testing, and diagnoses may not be made until 3 months after the first test. Using microarray analysis as the first step will save time and money and will give children with GDD and their families a better overall experience.

Reference

Trakadis, Y. and Shevell, M. (2011). Microarray as a first genetic test in global developmental delay: a cost-effectiveness analysis. *Developmental Medicine and Child Neurology, 53*, 994-999.