Special report: chromosomal microarray for the genetic evaluation of patients with global developmental delay, intellectual disability, and autism spectrum disorder

BlueCross BlueShield Association

Record Status
This is a bibliographic record of a published health technology assessment. No evaluation of the quality of this assessment has been made for the HTA database.

Citation

Authors' objectives
The objective of this Special Report is to summarize the evidence on chromosomal microarray testing to identify CNVs in children with GDD, ID, and ASD.

Authors' conclusions
The ability to detect pathogenic CNVs underlying GDD/ID and ASD is improving. This improvement is likely due to higher CMA resolution along with increasingly extensive data concerning CNV pathogenicity and associated phenotypes and availability of those data. Professional societies have recommended CMA testing as first-line evaluation when genetic evaluation is desired as opposed to first obtaining a karyotype. Data supporting analytic validity are readily available only for the Affymetrix CytoScan® Dx assay, but laboratories meeting CLIA standards but using other platforms would be expected to achieve adequate technical performance. There is consistent evidence that the diagnostic yield obtained from CMA testing is higher than with karyotyping in children with GDD/ID or ASD, with or without congenital anomalies. Establishing the pathogenicity of detected CNVs relies on evidence, informatics, and genetics expertise. A particular challenge when considering the evidence and methods used to determine variant pathogenicity is that, outside the more common syndromes, diagnoses include a large number of rare disorders a clinician, even a specialist, might not encounter during a lifetime. Identifying a pathogenic variant can: (1) impact the search for a diagnosis, (2) inform follow-up that can benefit a child by reducing morbidity, and (3) affect reproductive planning for parents and potentially the affected patient. Finally, we were unable to identify case reports of incorrect diagnoses; how often they might occur is unclear. For families desiring a genetic diagnosis and what might follow from one, CMA testing can establish a diagnosis more often than other approaches such as karyotyping. Still, other assays identify genetic alterations not detected by an array. The complexities of CMA testing, interpretation, understanding its limitations, and the potential implications requires that testing is obtained by clinicians with genetic expertise, that families receive genetic counseling, and that testing be performed in laboratories meeting recommended molecular pathology standards.

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