CLINICAL REPORT Guidance for the Clinician in Rendering Pediatric Care

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Genetic Evaluation of the Child With Intellectual Disability or Global Developmental Delay: Clinical Report

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Genetic neurodevelopmental disorders are common in the pediatric population, and establishing a specific diagnosis early provides multiple benefits including prognostication, surveillance for disorder-related complications, accurate recurrence risk, and specific management. This report provides an approach to the genetic evaluation of developmental delay/intellectual disability for the general pediatrician. When possible, genetic testing should be selected by phenotype, and typical distinguishing clinical features to facilitate this are presented. If a specific disorder or group of disorders cannot be ascertained by phenotype, an agnostic (or hypothesis-free) approach is utilized. Recommendations are provided for this agnostic approach based on diagnostic yield and also practical considerations such as test complexity and impact on management. The general guidance in this report for genetic testing does not preclude further evaluation by relevant subspecialists as necessary, including neurologists, developmental pediatricians, and clinical geneticists.

INTRODUCTION

Global developmental delay (GDD)/intellectual disability (ID) has an estimated global prevalence of 1% to 3%.[1] Further, in pediatric primary care, neurodevelopmental disorders are the most common chronic medical condition, with a combined prevalence of approximately 17%.[2] Neurodevelopmental disorders have a considerable impact at individual, family, and societal levels. Global developmental delay is defined in the *Diagnostic and Statistical Manual of Mental Disorders, 5th Edition* (DSM-5) as the failure to meet expected developmental milestones in several areas of intellectual functioning in an individual younger than 5 years who

abstract

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Drs Rodan and Stoler prepared and revised the manuscript. Drs Chen and Geleske revised the manuscript.

FINANCIAL/CONFLICT OF INTEREST DISCLOSURE: The authors have indicated they have no potential conflicts of interest to disclose.

To cite: Rodan LH, Stoler J, Chen E, et al; American Academy of Pediatrics, Council on Genetics. Genetic Evaluation of the Child With Intellectual Disability or Global Developmental Delay: Clinical Report. *Pediatrics*. 2025;156(1):e2025072219