

Chromosomal Microarray Analysis for Developmental Delay/Intellectual Disability, Autism Spectrum Disorder, or Congenital Anomalies

- I. Chromosomal microarray analysis for developmental delay, intellectual disability, autism spectrum disorder, or congenital anomalies (81228, 81229, S3870, 0209U) is considered **medically necessary** when:
 - A. The member has developmental delay and/or intellectual disability, excluding isolated speech/language delay (see below), **OR**
 - B. The member has autism spectrum disorder, **OR**
 - C. The member has multiple congenital anomalies not specific to a well-delineated genetic syndrome, **OR**
 - D. The member has short stature
- II. Chromosomal microarray analysis for developmental delay, intellectual disability, autism spectrum disorder, or congenital anomalies (81228, 81229, S3870, 0209U) is considered **investigational** for all other conditions of delayed development, including:
 - A. Isolated speech/language delay*

*See Background and Rationale section for more information about this exclusion.

NOTES AND DEFINITIONS

1. **Autism spectrum disorders:** is defined in the DSM V as persistent deficits in social communication and social interaction across multiple contexts, as manifested by the following, currently or by history:

- a. Deficits in social-emotional reciprocity, ranging, for example, from abnormal social approach and failure of normal back-and-forth conversation; to reduced sharing of interests, emotions, or affect; to failure to initiate or respond to social interactions.
 - b. Deficits in nonverbal communicative behaviors used for social interaction, ranging, for example, from poorly integrated verbal and nonverbal communication; to abnormalities in eye contact and body language or deficits in understanding and use of gestures; to a total lack of facial expressions and nonverbal communication.
 - c. Deficits in developing, maintaining, and understanding relationships, ranging, for example, from difficulties adjusting behavior to suit various social contexts; to difficulties in sharing imaginative play or in making friends; to absence of interest in peers.
2. **Congenital anomalies** according to ACMG are multiple anomalies not specific to a well-delineated genetic syndrome. These anomalies are structural or functional abnormalities usually evident at birth, or shortly thereafter, and can be consequential to an individual's life expectancy, health status, physical or social functioning, and typically require medical intervention.
3. **Developmental delay** is a slow-to-meet or not reaching milestones in one or more of the areas of development (communication, motor, cognition, social-emotional, or adaptive skills) in the expected way for a child's age
4. **Intellectual disability** (ID) is defined by the DSM V as:
 - a. Deficits in intellectual functions, such as reasoning, problem solving, planning, abstract thinking, judgment, academic learning, and learning from experience, confirmed by both clinical assessment and individualized, standardized intelligence testing.
 - b. Deficits in adaptive functioning that result in failure to meet developmental and sociocultural standards for personal independence and social responsibility. Without ongoing support, the adaptive deficits limit functioning in one or more activities of daily life, such as communication, social participation, and independent living, across multiple environments, such as home, school, work, and community.
 - c. Onset of intellectual and adaptive deficits during the developmental period.

REFERENCES

1. Moeschler JB, Shevell M; Committee on Genetics. Comprehensive evaluation of the child with intellectual disability or global developmental delays. *Pediatrics*. 2014;134(3):e903-e918. doi:10.1542/peds.2014-1839
2. Manning M, Hudgins L; Professional Practice and Guidelines Committee. Array-based technology and recommendations for utilization in medical genetics practice for detection of chromosomal abnormalities. *Genet Med*. 2010;12(11):742-745. doi:10.1097/GIM.0b013e3181f8baad
3. Manning M, Hudgins L; American College of Medical Genetics and Genomics (ACMG) Professional Practice and Guidelines Committee. Addendum: Array-based technology and recommendations for utilization in medical genetics practice for detection of chromosomal abnormalities [published online ahead of print, 2020 Jun 8]. *Genet Med*. 2020;10.1038/s41436-020-0848-8. doi:10.1038/s41436-020-0848-8
4. Mintz CS, Seaver LH, Irons M, Grimberg A, Lozano R, ACMG Professional Practice and Guidelines Committee. Focused Revision: ACMG practice resource: Genetic evaluation of short stature. *Genet Med*. 2021;23(5):813-815.