What is developmental delay?

Developmental milestones refer to skills or tasks that most children are able to accomplish by a specific age. Developmental delay is when a child does not meet developmental milestones in one or more areas. This generally refers to situations where someone is significantly and continually delayed, and not to those who are temporarily behind. A child is diagnosed with developmental delay after an assessment that includes evaluation of physical (fine motor skills, gross motor skills), cognitive (intellectual abilities), communication (speech and language), social or emotional development (social skills, emotional control), and adaptive (self-care) skills. The term developmental delay is used in early childhood (before school age or younger than age 5).

Some children will have delays in multiple areas. Children with significant delays in two or more areas are diagnosed with global developmental delay (GDD). Those with GDD are more likely to be diagnosed with intellectual disability (ID) later in childhood. However, not all children with GDD will have ID.

What happens once my child has been diagnosed with developmental delay?

If there are concerns that your child has developmental delays, their primary care physician or pediatrician may recommend further evaluation. This should involve a comprehensive review of your child's medical history, family history, physical and neurological exam, and may include a referral to meet with a genetics specialist.

What causes developmental delay?

Developmental delays may be caused by multiple factors, but for many the cause is unknown. Factors that can contribute include both genetic and environmental (nongenetic) factors (called multifactorial). Nongenetic factors can be things like exposures during the pregnancy, pregnancy or birth complications, poor nutrition, or infections. The chance that there is an underlying genetic condition is increased in those who have GDD and/or have other features or health problems, such as certain physical features, birth defects, hearing loss, or seizures.

What type of genetic testing may be recommended?

For children who have developmental delays, genetic testing may not always be recommended. The decision to proceed with testing may be based on the degree of delays that someone has, and whether they have other features or health problems. For some

children, the decision may be made to see how they grow and develop over time, and if there are continued concerns in the future genetic testing may be done.

There are multiple types of tests that may be recommended for those who have developmental delays. The most common include chromosome microarray and fragile X testing. The chromosome microarray is used to identify small missing or extra pieces of chromosome material, and can identify or rule out several hundred known genetic syndromes. Fragile X is a specific condition that can cause developmental delays along with other health issues. Other genetic testing may include single gene testing, multigene panels, or whole exome sequencing, but how useful additional testing like this would be depends on the individual and their specific health concerns.

Click here to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.

Additional Resources

Early Intervention

Language and Speech disorders

Learning Disorders

Developmental milestones