

From their first smile to their first steps and everything beyond, watching a child grow and develop is simply amazing. There are a lot of factors that can influence a child's development. These factors are often a delicate balance between nature and nurture, and can include such things as genetics, family, relationships, experiences, culture, education, nutrition, and much more. Here we are going to focus on the genetics of child development, the nature side of that equation.

Any expecting parents can tell you they have given quite a bit of thought to who their baby might look like. Will the baby have dad's chin? Or mom's eyes? We know traits like these are inherited within families: we look for them and we talk about them.

The individual instructions, or genes, that determine our traits and tell our bodies how to grow and function, are located on our chromosomes (for more background, visit Genetics 101). We are not all the same and not all children will follow the same path developmentally or medically, due in part to the complex interactions between our genes, and between our genes and environment. Some individuals may have differences that impact their development that cause difficulties in how they function day-to-day. These are called developmental disorders, and may include mental and/or physical difficulties. Examples include developmental delays, intellectual disability, autism spectrum disorder, hearing loss, vision impairment, and many others. According to the Centers for Disease Control (CDC), about 1 in 6 (~17%) children in the United States have a developmental disorder.

If you have concerns about your child's development, their primary care provider and your school district are often the first place to start asking questions. Many families of young children (under age 3) are surprised to learn their school district offers evaluation and early intervention services at no charge.

When a baby or child begins showing signs of differences from their family background or their peers, their primary care provider may refer them for a developmental assessment. These assessments typically include physical development (fine motor skills, gross motor skills), cognitive development (intellectual abilities), communication development (speech and language), social or emotional development (social skills, emotional control), and adaptive development (self-care skills). If there are developmental concerns noted, a child's primary care provider may also refer them to see a specialist, such as a genetics professional.

Below is a list of common reasons why a child may be referred for genetics evaluation:

1. Developmental delays



- 2. Intellectual disability
- 3. Autism spectrum disorder
- 4. Unusual growth (poor growth, overgrowth, or asymmetric growth)
- 5. Multiple physical differences noted at birth (also called birth defects)
- 6. Facial features that do not appear similar to relatives
- 7. Abnormal labs (blood or urine testing) or imaging studies (like <u>x-rays</u> or <u>ultrasounds</u>)
- 8. <u>Congenital (from birth) hearing loss</u>

A genetics evaluation involves a review of the child's medical history, family history, and detailed physical examination. Based on this evaluation, genetic testing may be recommended. The results that come from genetic testing may provide your family with helpful information. An informative or 'positive' test result may help explain why a child has developmental differences, provide insight into additional or future medical needs, provide your family with the likelihood for future children to have similar symptoms, and provide access to a community of families/children with the same diagnosis. An uninformative or negative test result may rule out certain genetic conditions and may mean additional testing is recommended

Click on the links below to learn more about different types of genetic testing that are available.

- Chromosome microarray
- Single gene testing
- Multigene Panel/NGS
- Whole Genome or Whole Exome sequencing

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

Additional Resources

Center for Disease Control and Prevention

Typical Child Development (Birth to three)

Early Intervention

American Academy of Pediatrics (Individualized Education Program)