



What is intellectual disability?

Intellectual disability (ID) is a developmental disorder where an individual has significant difficulties with intellect and independent functioning that is identified before they turn 18 years old. In the past, intellectual disability has also been called mental retardation and cognitive disability. ID is a spectrum in which individuals may range from being more mildly to more profoundly affected in their abilities. The diagnosis of ID is typically made following the use of a standardized measurements of skills. About 1-3% of the general population is affected by ID.

What causes intellectual disability (ID)?

Multiple factors can contribute to someone developing ID, and can include both genetic and environmental (nongenetic) influences (called [multifactorial](#)). About 40% of those with ID, and up to about 50% of those with moderate to severe ID, will have an underlying genetic cause. Some nongenetic causes of ID include prenatal exposures (e.g. alcohol), infection during pregnancy or in childhood, birth complications, and brain injury or trauma.

For those who have an underlying genetic condition as the cause of their ID, they may also have a chromosome abnormality or single gene disorder (such as [PTEN Hamartoma Tumor syndrome](#)) which can involve other health issues. The likelihood of an underlying genetic condition may be impacted by whether a child has other features or health problems, such as distinctive physical features, birth defects, seizures, etc. This is often referred to as “syndromic” ID, and is more likely to have a genetic cause.

What type of genetic testing may be recommended?

There are multiple types of tests that may be recommended for those who have ID. The most common is the [chromosome microarray](#). 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. This testing has a detection rate (the chance for the testing to come back positive) of about 10-30% in those with ID.

Further genetic testing that may be considered includes single gene testing, [multigene panels](#), or [whole exome sequencing](#) to identify a single gene disorder. Examples of single gene testing include [fragile X](#) (accounts for less than 5% of those with ID) or [MECP2](#) gene testing (accounts for less than 2-3% of females with [GDD/ID](#)). Through the use of [multigene panels](#) and whole exome sequencing, an underlying genetic cause is found in about 30-40% of those with ID.



Other testing that may be recommended includes biochemical screening, which involves measuring the levels of different substances in the blood or urine. These tests are used to look for metabolic conditions that can also cause ID. These type of conditions account for less than 5% of those with ID.

Click [here](#) to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.

Additional Resources

[The Arc](#)

[The American Psychiatric Association](#)

[American Academy of Pediatrics](#)

[American Academy of Pediatrics](#) (Individualized Education Program)