

PEDIATRIC GLOBAL DELAY

Define&Decide

Harnessing
genetics
for a **clear**
diagnosis



MEDICOVER
GENETICS

WHAT IS **PEDIATRIC GLOBAL DELAY** Define&Decide?

DEFINE

Global developmental delay and intellectual disability (GDD/ID) affect up to 3% of children <5 years old and is defined as a delay in ≥ 2 developmental domains*. Up to 40% of GDD/ID cases are caused by genetic factors, including chromosomal abnormalities in 25% of cases, and monogenic disorders in up to 10% of cases. Up to two-thirds of children with GDD do not have a single group of symptoms that can point towards a specific diagnosis; therefore, several genetic tests are often required to define the cause of GDD/ID.

Our tests combine chromosomal analyses and comprehensive gene panels associated with many different disorders with overlapping features.

DECIDE

Having a diagnosis can help you decide on early management and treatment options and identify associated medical risks, thereby improving the patient's clinical outcome and preventing further complications. Our genetic counselling can help guide management options and reproductive decisions based on recurrence risks.

WHO COULD **BENEFIT** FROM THIS TEST?



Children <5 years with a significant delay in ≥ 2 developmental domains*

Children with an autism spectrum disorder

Children with dysmorphic features

**Developmental domains include physical, cognitive, speech/language, social and emotional*

WHY **RECOMMEND** TESTING?

Genetic information can help:

- Decide on early management and treatment options
- Identify associated medical risks, thereby improving the patient's clinical outcome and preventing further complications
- Guide reproductive decisions based on recurrence risks

OUR TESTS

We offer advanced genetic testing options including microarray CGH, gene panels, and Whole Exome Sequencing (WES) analysis. The tests were compiled based on their relevance to the disorders tested.

MICROARRAY CGH

GENE PANELS

WHOLE EXOME SEQUENCING

Fragile X syndrome analysis is available upon request.

Microarray comparative genomic hybridization (microarray CGH)

Used for genome-wide screening of deletions and duplications.

- Does not require prior knowledge of precise genetic aberrations
- Will not detect chromosomal structural changes that do not result in deletions/duplications, such as translocations or inversions, ring chromosomes or low-level mosaicism

Gene panels

Our gene panels are designed to identify disorders characterized by overlapping phenotypic features, facilitating a more accurate diagnosis. These panels target specific genes known to be associated with GDD/ID.

- Autism
- Coffin-Siris syndrome
- Congenital disorders of glycosylation
- Cornelia de Lange syndrome
- Developmental disorders
- CHARGE syndrome
- Coffin-Lowry syndrome
- Fragile X syndrome
- Glycosylphosphatidylinositol biosynthesis defect
- Hydrops fetalis
- Kabuki syndrome
- Macrocephaly
- MECP2 duplication syndrome
- Microcephalic osteodysplastic primordial dwarfism
- Microcephalies, primary, AR
- Mowat-Wilson syndrome
- Neurotransmitter disorders, pediatric
- Overgrowth syndromes
- Pitt-Hopkins syndrome
- Rett syndrome
- Rett syndrome & Rett syndrome-like disorders
- Robinow syndrome
- Rubinstein-Taybi syndrome
- Sotos syndrome
- Weaver syndrome

Whole exome sequencing (WES)

Comprehensive test that examines the coding regions (exons) of the human genome.

- Can identify genetic variations responsible for a wide range of inherited disorders
- Three testing options:

Trio WES

patient and 2 biological parents

highest diagnostic yield

Duo WES

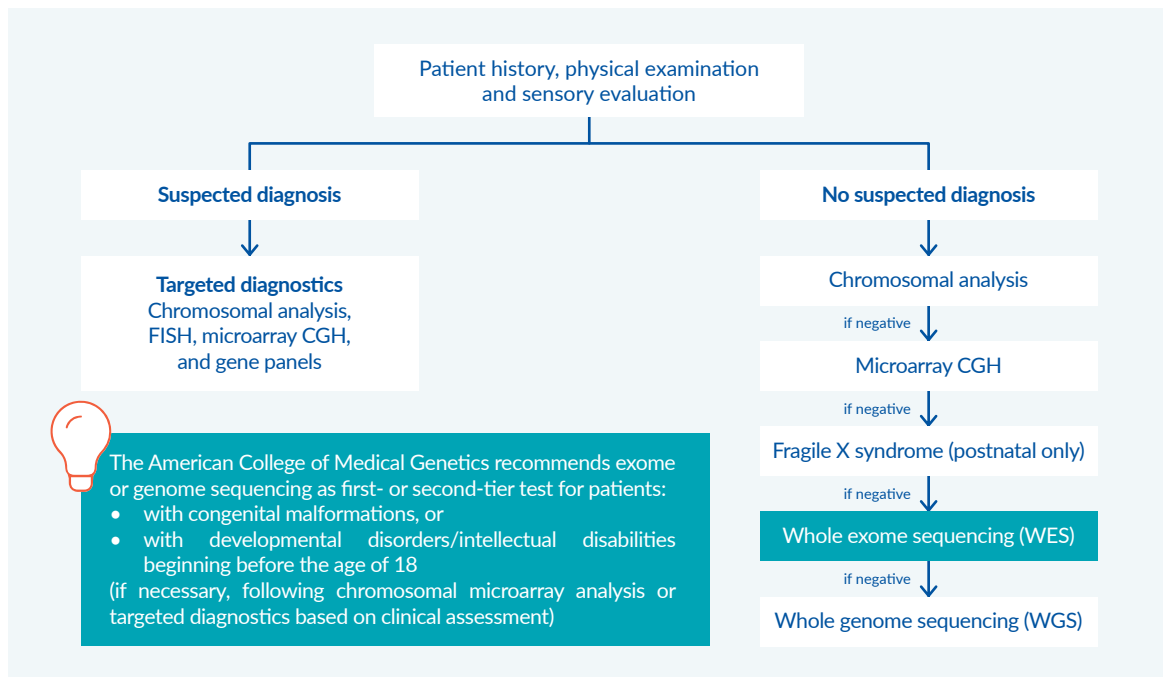
patient and 1 biological parent

Single WES

patient only

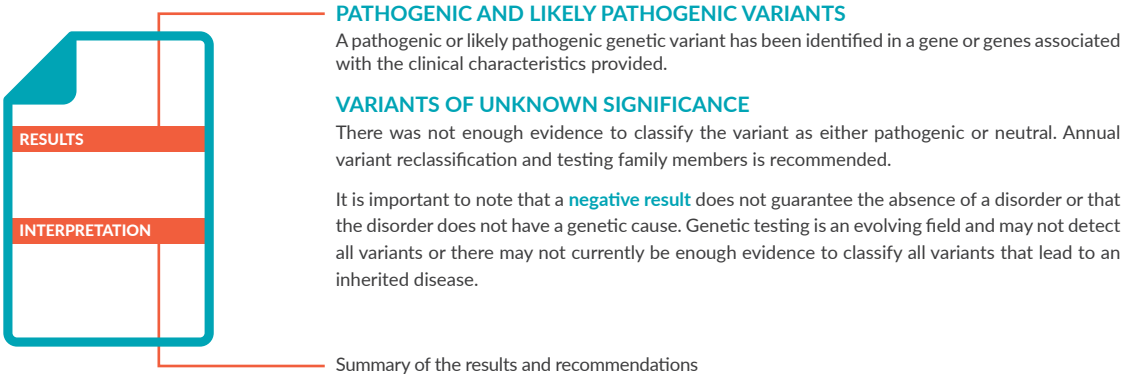
DIAGNOSTIC ALGORITHM

Tests should be chosen according to the diagnostic algorithms recommended by international societies.



WHAT ARE THE POSSIBLE OUTCOMES OF THE TEST?

A molecular genetic diagnostic report outlining the results of the sequencing analysis is provided. Changes in DNA sequences (variants) can be detrimental and lead to a disorder causing GDD/ID. We will report on the following types of variants:



Interpretation of the molecular genetic results relies on an accurate clinical picture of the patients

TECHNICAL DETAILS

MICROARRAY CGH

- Labeled and hybridized with reference DNA on a 180K-Array
- Washed, scanned, and analyzed using specialized software
- Mutations covered: deletions and duplications
- Functional resolution: 50kb

NEXT GENERATION SEQUENCING

- Used for gene panels and WES
- Performed on exons and conserved intronic regions
- Mutations covered: SNVs, small INDELs and CNVs
- Human reference genome: GRCh38
- Median read depth: 100x
- Coverage: >97-99% over 20x
- Variant classification follows ACMG guidelines

HOW TO ORDER?



Recommend Pediatric Global Delay analysis to your patient



Collect the sample(s)



Send the sample(s) to **Medicover Genetics**



The sample(s) will be analyzed at **Medicover Genetics** laboratories



Results will be sent to you

MORE QUESTIONS?

If you have additional questions or concerns, please contact us at info.genetics@medicover.com



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