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 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.

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

Our analyses

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.

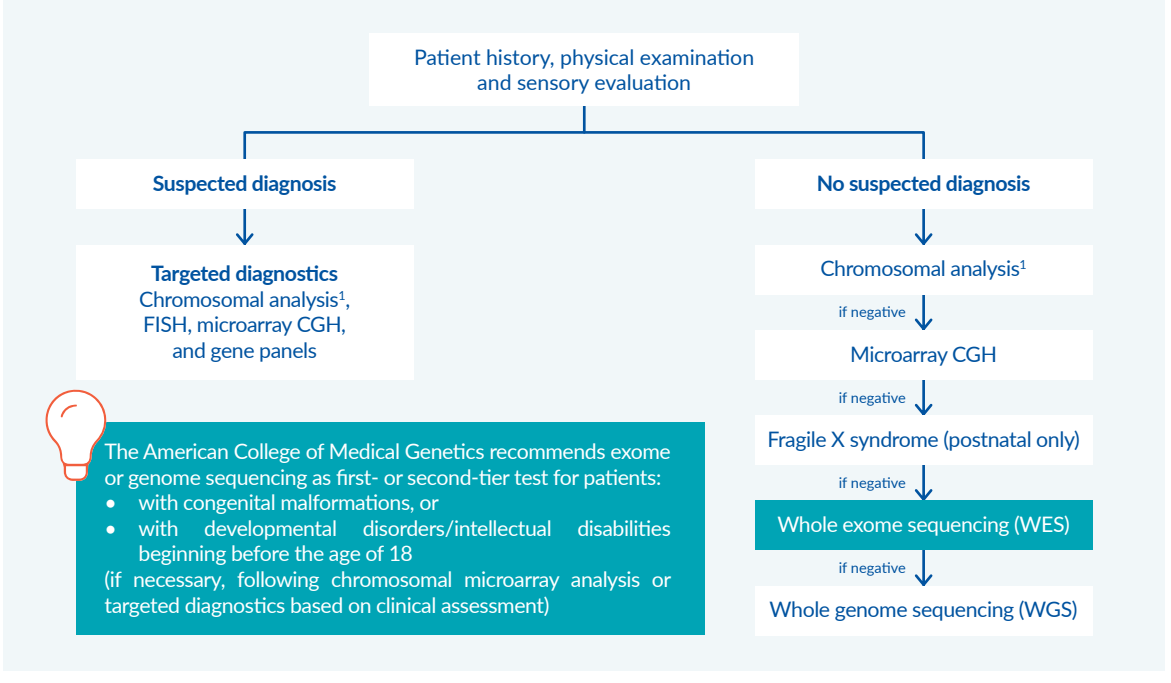
- Can identify genetic variations responsible for a wide range of inherited disorders
- Three main testing options:

Trio WES	Duo WES	Single WES
patient and 2 biological parents	patient and 1 biological parent	patient only

highest diagnostic yield

DIAGNOSTIC **ALGORITHM**

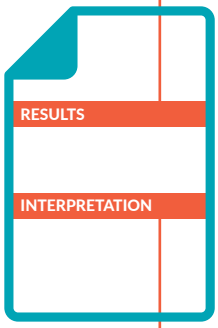
Tests should be chosen according to the diagnostic algorithms recommended by international societies.



[1]*Availability limited to Germany at this time*

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:



PATHOGENIC AND LIKELY PATHOGENIC VARIANTS

A pathogenic or likely pathogenic genetic variant has been identified in a gene or genes associated with the clinical characteristics provided.

VARIANTS OF UNKNOWN SIGNIFICANCE

There was not enough evidence to classify the variant as either pathogenic or neutral. Annual variant reclassification and testing family members is recommended.

It is important to note that a **negative result** does not guarantee the absence of a disorder or that the disorder does not have a genetic cause. Genetic testing is an evolving field and may not detect all variants or there may not currently be enough evidence to classify all variants that lead to an inherited disease.

Summary of the results and recommendations

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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