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 and can occur in isolation or accompanied by other symptoms including malformations and neurological disorders. Children with GDD/ID exhibit mixed and diverse symptoms, and up to two-thirds do not have a single group of symptoms that can point towards a specific diagnosis. As a result, many patients undergo a long diagnostic journey before necessary genetic tests are performed to define the cause of GDD/ID.

Our tests combine chromosomal analyses and (comprehensive) gene panels associated with many different disorders with overlapping features, providing a diagnostic solution for children with GDD/ID.

DECIDE

Having a diagnosis can help you decide on a management plan or treatment options for your child. Our genetic counselling offers information regarding the diagnosis, identifies associated medical risks, and provides a long-term prognosis, thereby improving your child's clinical outcome and may help prevent further complications.

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*

IMPORTANCE OF GETTING TESTED

Children with a GDD/ID disorder often require lifelong support, which can have a profound effect on their lives and that of their family. A timely diagnosis is crucial for therapeutic intervention and the best outcome for your child. Identifying the cause can provide a prognosis, refine treatment options, evaluate recurrence risks and provide closure to the diagnostic journey. In turn, this can improve your mental health and that of your child, while validating your concerns and empowering you to advocate for your child.

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 (loss of genetic material) and duplications (gain of genetic material)

- 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.

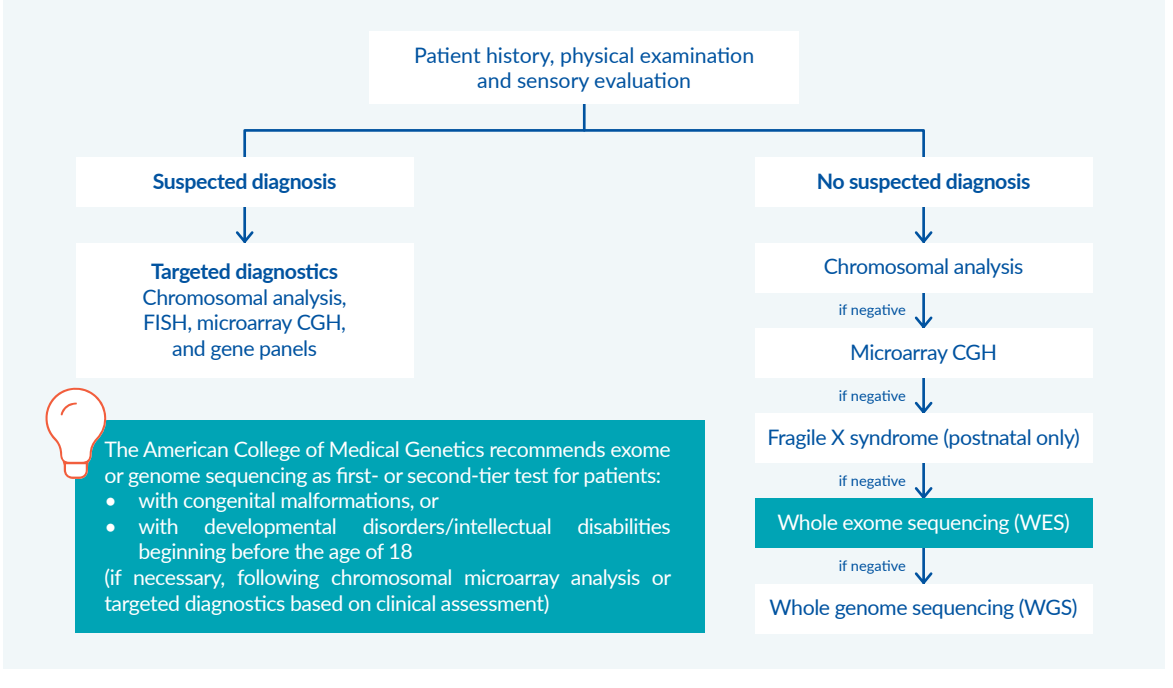
- Can identify genetic variations responsible for a wide range of inherited disorders
- Three testing options:

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

highest diagnostic yield

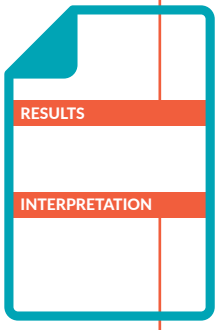
DIAGNOSTIC **ALGORITHM**

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



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

MEDICAL **GENETIC COUNSELLING**

Medical genetic counselling is an essential part of a genetic testing journey that we offer before and after testing. Genetic counsellors will obtain a detailed family history, explain the method of testing that will be used, its risks and benefits, the limitations of the diagnosis, and advise you on the consequences of the results including management options and recurrence risk. The goal of counselling is to provide you with a greater understanding of the results and the ability to make more informed choices for your child.

Availability of genetic counselling services may vary by country. Please contact us to check for more information on access in your region.

HOW TO **ORDER**?

Visit a physician or one of our medical geneticists to choose the right test for you

The sample is sent to **Medicover Genetics**

Discuss the medical report with your physician

Arrange sample collection at your nearest blood drawing point (accepted samples: EDTA blood, buccal swab)

Sequencing is performed in our accredited laboratory in Germany

MORE **QUESTIONS**?

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

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