MEDICOVER
GENETICS

PEDIATRIC GLOBAL DELAY

Define&Decide

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Patient Information

WHAT IS 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.

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

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

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.

MEDICAL GENETIC COUNSELLING

Medical genetic counselling is an essential part of a genetic testing journey that we offer before and after testing. Our 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.

DIAGNOSTIC PROCESS

STEP 1

Patient history, **physical examination** and **sensory evaluation** should be conducted for each child with suspected GDD/ID

STEP 2

Following a clinical evaluation, **genetic counselling** is recommended with one of our counsellors

STEP 3

Molecular genetic analysis of the patient's genome

WE OFFER TWO OPTIONS TO TEST FOR GDD/ID:

OPTION 1: Stepwise Analysis

First, a genome-wide screen for **deletions/duplications** is performed. If none are detected, another **genetic counselling session** is conducted and one of our **gene panels** will be recommended by our genetic counsellor.

OPTION 2: Simultaneous Analysis

Screening for **deletions/duplications** and **gene panel sequencing** are performed **at the same time**. Performing both steps simultaneously saves time and resources for the patient.

STEP 4

All cases are finalized with a **medical report** and **genetic counselling**

OUR TESTS

SCREENING OF DELETIONS, DUPLICATIONS AND ANEUPLOIDIES

Microarray comparative genomic hybridization (microarray CGH) is used for genome-wide screening of deletions (loss of genetic material) and duplications (gain of genetic material) and does not require prior knowledge of precise genetic aberrations. This method 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

- 63 genes — Autism Spectrum Disorders
- 43 genes — Brain Malformations, Comprehensive
- 9 genes — Brain Malformations, Lissencephaly
- 19 genes — Brain Malformations, Pontocerebellar Hypoplasia
- 8 genes — Brain Malformations, Tubulinopathies
- 10 genes — Coffin-Siris Syndrome
- 43 genes — Congenital Disorders Of Glycosylation
- 7 genes — Cornelia De Lange Syndrome
- 21 genes — GPI Anchor Deficiency
- 179 genes — Intellectual Disability
- 66 genes — Macrocephaly
- 30 genes — Microcephaly
- 11 genes — Overgrowth Syndromes
- 14 genes — Pediatric Neurotransmitter Disorders
- 20 genes — RASopathies, Comprehensive
- 3 genes — Rett Syndrome
- 22 genes — Rett Syndrome And Related Disorders

- Fragile X syndrome analysis is available upon request
- Interpretation of the molecular genetic results relies on having an accurate clinical picture of the patient

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: the genetic cause of the observed symptoms has been identified and may help determine the right treatment and management plan.

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 may not be able to detect all variants that lead to an inherited disease.

DEVELOPMENTAL DOMAIN SYMPTOMS

GROSS AND FINE MOTOR: Delayed ability to sit, crawl or walk • Delayed ability to jump, run and climb • Inability to grasp objects • Inability to hold utensils, work with objects, and draw
SPEECH AND LANGUAGE: Difficulty speaking or speaking late • Difficulty understanding language • Inability to express thoughts
COGNITION: Lack of curiosity • Short attention span and easily distracted • Inability to remember things • Inability to connect actions with consequences • Difficulty with problem-solving or logical thinking
PERSONAL AND SOCIAL DEVELOPMENT: Difficulty communicating or socializing with others • Inability to express and control emotions • Lower than average IQ test scores • Showing repetitive and restricted behavior • Showing extreme behavior (unusually fearful, aggressive, shy or sad)
ACTIVITIES OF DAILY LIVING: Inability to do everyday tasks like getting dressed, eating, brushing teeth, washing hands or going to the bathroom without help

Please note that symptoms vary in type and severity between children and that not all symptoms are listed for each developmental domain. Children should be assessed by your physician and/or genetic counsellor. *Adapted from https://www.cdc.gov/ncbddd/actearly/milestones*

WHY US

- A network of laboratories and medical institutions makes us **a leader in genetic testing** in Germany with foundations dating back to 1998
- A clinical team comprised of scientists, physicians and medical geneticists, several with **>20 years of experience** in genetic testing, assuring meaningful and comprehensive genetic tests
- **Up-to-date diagnostic algorithms**
- Expertise in gene variant analysis ensuring **"no variant left behind"**
- Cutting-edge technology in sequencing and laboratory methods allows for **short turnaround times**
- **Quality** assessed by several certifying bodies, including EFI, DIN EN ISO 15189 accreditation for medical laboratories, DIN EN ISO/IEC 17025 accreditation for testing and calibration laboratories and a generally valid GMP (Good Medical Practice) certificate
- **Data privacy** is your right and our priority

HOW TO GET THIS TEST ORDERED

A physician or medical geneticist chooses the right test

A sample is collected at the nearest blood drawing point

Analysis is performed in our laboratory

A medical report is delivered

Genetic counselling is available upon request

Step 3-4 takes 15-25 working days

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Contact info

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