

## Joubert Syndrome Panel

The DBGen Joubert syndrome panel includes a genetic study of 45 genes known to cause the disease. This panel is part of the Complete Panel of Retinal Dystrophies and other eye diseases.

### ABOUT JOUBERT SYNDROME

Joubert syndrome exhibits mainly autosomal recessive pattern of inheritance. This disorder is a ciliopathy that affects different areas of the brain. It is characterized by a loss of muscle tone, truncal ataxia, dysplasia and retinitis pigmentosa, as well as other symptoms. Its prevalence is between 1:80,000 and 1:100,000.

#### **PATHOLOGIES**

The panel includes the genes most often responsible for the following disorders:

- >> COACH syndrome
- >> **Joubert syndrome**
- >> Meckel syndrome
- >> Nephronophthisis
- >> Senior-Loken syndrome

#### **GENES ANALYZED**

AH11, ANKS6, ARL13B, ARL6, B9D1, B9D2, C5orf42, CC2D2A, CEP164, CEP290, CEP41, CEP83, CSPP1, GLIS2, IFT27, IFT81, INPP5E, INVS, IQCB1, LZTFL1, MKKS, MKS1, NEK8, NPHP1, NPHP3, NPHP4, OFD1, PDE6D, RPGR, RPGRIP1L, SDCCAG8, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, TTC8, WDPCP, WDR19, ZNF423

Non-coding regions included:  
CEP290 c.2991+1655A>G, OFD1 c.935+706A>G

### PRICE

From €825. Please contact us to know the options that best suit your needs.

### RESULTS

A detailed genetic report that includes the genetic variants iden-

tified and genetic counseling will be provided. Supporting information will be exhaustive based on bibliographical studies and database analyses and, especially, on our 25 years of experience researching the genetics of hereditary eye diseases.

The test will be performed once payment is made and the signed informed consent and the sample are received. The report will be delivered 12 to 14 weeks after the above conditions are satisfied.

### METHODOLOGY

The diagnostic strategy relies on the automated sequencing of DNA on Illumina HiSeq 2000 sequencers that are specially designed for this kind of high-performance analysis. Our panels have been designed to prioritize the genomic regions associated with the hereditary eye diseases indicated in this text.

The likely pathogenic nucleotide variants are verified using Sanger sequencing. We check that their frequency in the control population is below 1% and that they meet the pathogenicity predictions as per established bioinformatics algorithms (SIFT, LRT, MutationTaster, PolyPhen2, CADD and NetGene2).

### RECOMMENDED FOR

This test is recommended when the clinical diagnosis indicates one of the pathologies previously listed and when the clinical condition is clearly defined.

This panel offers a high diagnostic performance because it includes all of the genes known to cause these diseases and because the method used allows identifying certain structural genomic alterations that are difficult to detect with other test types.