ABOUT RETINAL DYSTROPHIES AND OTHER EYE DISEASES

This is a set of degenerative diseases of the retina and optic nerve that exhibit a wide genetic and clinical heterogeneity. These diseases can be stationary or progressive from an early age. They can also occur in isolation or in syndromic forms. The inheritance pattern can be autosomal dominant, autosomal recessive or X-linked. Over 300 genes are known to cause these types of disorders, most of them affecting the expression of proteins involved in phototransduction, the structure of photoreceptors and the cellular pathways needed for viability and function in retinal cell. Moreover, mutations in the same gene can cause different pathologies.

PATHOLOGIES

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

- Achromatopsia
- Albinism
- Alström syndrome
- Aniridia
- Axenfeld-Rieger syndrome
- Bardet-Biedl syndrome
- Bestrophinopathy
- Bietti crystalline dystrophy
- Bosch-Boonstra-Schaaf opticotrophy syndrome
- Bothnia retinal dystrophy
- Bradyopsia
- Central areolar choroidal dystrophy
- Cho-roideremia
- COACH syndrome
- Cohen syndrome
- Cone-rod dystrophy
- Congenital stationary night blindness
- Costeff optic atrophy syndrome
- Doyne honeycomb degeneration of retina
- Enhanced S-cone syndrome
- Exudative vitreoretinopathy
- Familial benign fleck retina
- Fundus Albinopunctatus
- Glaucoma
- Gyrate atrophy of choroid and retina
- Hermansky-Pudlak syndrome
- Jalili syndrome
- Joubert syndrome
- Knobloch syndrome
- Leber congenital amaurosis
- Mainzer-Saldino syndrome
- Mckusick-Kaufman syndrome
- Meckel syndrome
- Mevalonate kinase deficiency
- Nephronophthisis
- Norrie disease
- Oculocutaneous albinism
- Oguchi disease
- Open-angle glaucoma
- Optic atrophy
- Optic nerve hypoplasia
- Peters anomaly
- Pigmented paravenous chorioretinal atrophy
- Refsum disease
- Retinitis pigmentosa
- Retinoschisis
- Rhegmatogenous retinal detachment
- Senior-Loken syndrome
- Sorsby Fundus Dystrophy
- Stargardt disease
- Stickler syndrome
- Usher syndrome
- Vitelliform macular dystrophy type I
- Vitelliform macular dystrophy type II
- Vitreoretinochoroidopathy
- Waardenburg syndrome
- Wagner syndrome

RESULTS

A detailed genetic report that includes the genetic variants identified 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.

PRICE

From €950. Please contact us to know the op-
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 a disease of the retina or optic nerve in both syndromic and non-syndromic cases, and especially when the clinical conditions are not clearly defined.

This panel offers a very high diagnostic yield (>79%) as it includes a high number of causative genes and because the method used allows identifying some structural genomic alterations difficult to characterize with other test types.

GENES ANALYZED
Simultaneously sequence of all the coding regions (exons) of 346 genes

And 66 relevant intronic regions of 13 genes:

www.dbgen.com