

PGD for monogenic diseases



• If you have an inherited disease, we can help you have a healthy baby

[www.igenomix.com](http://www.igenomix.com)

**igenomix**  
PIONEERS IN REPRODUCTIVE GENETICS



IGENOMIX is a company that provides advanced reproductive genetics services.

Our broad experience and qualifications make us one of the global leaders in this field, and we offer couples effective solutions that are at increased risk to have a child with a hereditary genetic disease.

Our aim is to prevent the transmission of monogenic diseases, which are serious diseases caused by the alteration of one gene to offspring. To achieve this, we offer families accurate diagnosis of embryos performed by highly qualified experts using the latest advanced technology.

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## What are chromosomes and genes? How does a monogenic disease occur?

In a person, each cell contains chromosomes that were inherited from each parent, 23 from the father and 23 from the mother. Therefore, each person has two pairs of 23 chromosomes, or 46 total chromosomes.

Chromosomes are comprised of molecules called DNA. Our DNA is organized into small fragments called genes. When the function of the gene is altered by a change, called mutation, in the specific sequence a monogenic disease results.

These mutations can be transmitted in families from generation to generation, or can be a new change in an individual (de novo).

- ❖ When the function of the gene is altered by a change, called mutation, in the specific sequence a monogenic disease results

❖ PGD for monogenic diseases is an early genetic diagnosis of an embryo



## What is Preimplantation Genetic Diagnosis (PGD) for monogenic diseases?

To prevent transmission of genetic diseases, IGENOMIX recommends that Preimplantation Genetic Diagnostic (PGD) be performed.

PGD for monogenic diseases is an early genetic diagnosis of an embryo prior to transfer to the uterus and, therefore, prior to establishment of pregnancy. By analyzing DNA from each embryo, embryos without the mutation can be preferentially selected. These embryos can be transferred back into the woman's uterus.

PGD helps couples who have an increased risk for a genetic disease conceive a healthy pregnancy.

❖ To prevent transmission of genetic diseases, IGENOMIX recommends that Preimplantation Genetic Diagnostic (PGD) be performed



## ❖ In IGENOMIX, we can perform PGD for most monogenic diseases

This type of PGD is indicated for couples with a personal or high risk in the family for any monogenic disease which includes Cystic Fibrosis, Fragile X Syndrome, a muscular dystrophy or atrophy or Huntington disease.

In IGENOMIX, we can perform PGD for most monogenic diseases. However we have a panel of common diseases in which the PGD test is already developed.

### PANEL OF FREQUENT MONOGENIC DISEASES

- Autosomal Dominant Polycystic Kidney Disease
- Autosomal Recessive Polycystic Kidney Disease
- Becker muscular dystrophy
- Beta thalassaemia
- Congenital adrenal hyperplasia (gene CYP21A2)
- Cystic fibrosis
- Charcot-Marie-Tooth type 1A
- Duchenne muscular dystrophy
- Familial amyloid polyneuropathy
- Fragile X syndrome
- Hemophilia A (F8)
- Hemophilia B (F9)
- Huntington's disease
- Multiple endocrine neoplasia, type 2A
- Myotonic dystrophy (Steinert)
- RhD incompatibility
- Spinal muscular atrophy
- X-linked adrenoleukodystrophy

We also offer PGD for any other monogenic disease, which will require a personalized development

❖ Consult your physician for further information and assessment. For any questions, please contact us directly



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