

# Karyomapping

Preimplantation genetic diagnosis





## How does karyomapping differ from other PGD tests?

Until recently, PGD tests for disorders caused by inheritance of a defective gene had to be tailor-made for each couple. This required weeks or months of work by highly skilled scientists, meaning that costs were high and there was often a long wait before IVF treatment could begin. By contrast, karyomapping provides a test for PGD of almost any known gene defect with a rapid turnaround time. Since karyomapping uses thousands of markers per chromosome it is likely to be more accurate than current tests.

## Where is karyomapping available?

Karyomapping is available in a number of leading fertility clinics worldwide. If you are considering this type of treatment and would like to find out whether it is available at your chosen IVF clinic, please enquire with them directly as it is important that each couple has their individual circumstances assessed appropriately by a clinician.

# Who is karyomapping for?

You may know that you are a carrier of a genetic disorder because you already have an affected child, or you may be aware of a family history of the disorder and have had your DNA tested.

Karyomapping is a new technique that allows couples, known to be carriers or affected with an inherited condition, to avoid passing on that disorder to their offspring. The technique works by screening embryos for the disorder before implantation in the womb (uterus)—a technique called preimplantation genetic diagnosis, or PGD.

Using PGD greatly reduces the chance that a fetus will be affected by the genetic disorder and, consequently, it is much less likely that termination of a pregnancy will need to be considered, or that an affected child will be born.

## Why is IVF required?

In order to carry out PGD *in vitro* fertilization (IVF) is essential. This process involves collecting several eggs from the ovaries and fertilizing them outside the body (*in vitro*) to produce several embryos. Each of the embryos can then be tested to find out which are suitable for transfer.

Only the embryos that are predicted to be free of the genetic condition are transferred to the uterus and, consequently, any pregnancy that begins has a low risk of being affected by the disorder.

# How does karyomapping work?

A blood sample is taken from the father, the mother, and a close relative of known disease status (affected, unaffected or a carrier). In most cases, the relative tested is a child of the couple. This relative is referred to as 'the reference'.

Karyomapping looks at the chromosomes, the long rod-like structures that are found in cells and contain the genes. Karyomapping examines the chromosomes of the mother, father, and the reference at approximately 300,000 different points and finds a DNA fingerprint unique to the chromosome that carries the defective gene. It is then possible to test embryos produced using IVF for this fingerprint, revealing those that have inherited the affected chromosome.

If the fingerprint characteristic of the chromosome carrying the defective gene is not detected, then it can be inferred that the embryo has inherited normal copies of the gene and is therefore likely to be free of the disorder. Embryos of this type are good candidates for transfer to the mother's uterus (womb).

Ask your doctor  
if karyomapping is  
right for you.

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For more information on PGD, please visit:

**[www.chromosome-screening.org](http://www.chromosome-screening.org)**

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