Karyomapping:

A rapid PGD solution for single gene disorders.

How does karyomapping work?

Until now, PGD has been performed by examining the inheritance of short tandem repeats (STRs) adjacent to specific disease loci. Each STR test for a genetic disorder must be developed individually for each couple requesting the test, and for each genetic disorder. This workup requires weeks or months of work by highly skilled scientists, meaning that costs are high and there's often a long wait before treatment can begin. The workup and validation of each STR marker typically takes 3 to 6 months.

PGD of single gene disorders using karyomapping doesn't require the development of disease or patient specific tests, so it significantly reduces the time required. A typical turnaround time would be less than one week. Following a simple blood draw from the parents and a close relative of known disease status, e.g. an existing child, the method uses SNP genotyping. For each embryo, SNPs are used to identify the parental origin of chromosomes at all SNP loci.

Approximately 300,000 SNPs provide genome wide coverage, meaning that any single gene disorder can be screened for.*

Once the disease loci for the specific genetic disorder have been established, it can be determined whether the section of the DNA from the parent that carries the mutation for the genetic disorder matches the section of DNA in the embryo—if it does then it is inferred that the embryo also carries the mutation and is likely to be affected or a carrier of the mutation. This is done through comparing the DNA fingerprint with a relative of known disease status termed as a reference.

If the section of DNA from the parent that carries the mutation isn't seen in the embryo, then it's inferred that the embryo is unaffected and is likely to be without the disease-causing mutation and would therefore be a good candidate for transfer.

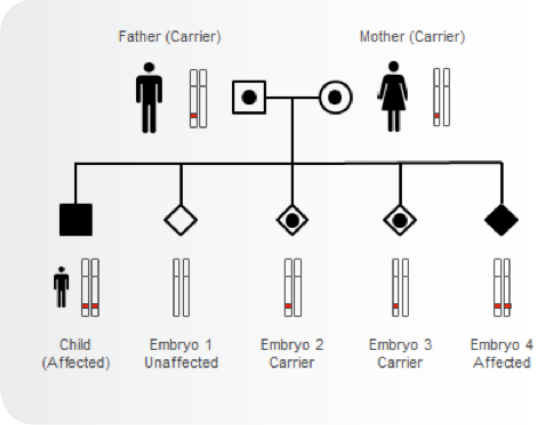
Single gene PGD

A couple may present when they know that one or both of them are a carrier of a genetic disorder, or someone in the family has the genetic disorder, or because they already have an affected child.

Karyomapping is a new technique that allows these couples to avoid passing that disorder on to their offspring[1]. The technique works by screening embryos before implantation in the 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.

Karyomapping: PGD Screening for single gene disorders



Benefits of karyomapping

- Ultra-rapid workup time compared to current STR technology
- No disease-specific or patient-specific workup required
- Available for all single gene disorders*
- No change in preparation needed by IVF clinic
- IVF clinics can access karyomapping from specialist genetic testing centers

How to access karyomapping?

Samples are biopsied locally at the IVF clinic and then karyomapping is performed at a specialist genetic testing laboratory—a reference lab. Couples do not need to travel to the testing site, but can be kept informed by their chosen IVF clinic. The IVF clinic receives a report from reference lab detailing the disease status of each embryo in order to inform embryo selection.

*Karyomapping must only be used for PGD conditions licensed by local laws.

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For more information on PGD, please visit:
www.chromosome-screening.org

Sources:

1. Handyside *et al*. (2010). Karyomapping: A Universal Method for Genome Wide Analysis of Genetic Disease Based on Mapping Crossovers between Parental Haplotypes. *J Med Genet.* Oct;47(10):651-8.