What genetic conditions can be detected by PGD?

It is possible to perform PGD for any genetic alteration, autosomal dominant, recessive or X-linked, given that there is an identifiable gene. For trinucleotide repeat disorders (like fragile X syndrome, Huntington's disease, etc.) specific detection techniques for these alterations are performed.

BIOARRAY ADDITIONALLY PROVIDES THE CHROMOSOMAL PROFILING RESULT (PGS) TO COUPLES UNDERGOING PGD FOR A SINGLE-GENE DISORDER

Bioarray analyzes more than 700 genes associated to single-gene disorders like:

- Alport syndrome
- Beta thalassemia
- Dravet syndrome
- Hemophilia A/B
- Marfan syndrome
- Spinal Muscular Atrophy
- Charcot-Marie-Tooth
- Cystic fibrosis
- Lynch syndrome
- Polycystic kidney disease

... CONSULT WITH BIOARRAY FOR ANY OTHER GENETIC DISORDER, INCLUDING MITOCHONDRIAL DISEASES

NGS for PGD: Bioarray´s validated technique

Bioarray launches the new PGD using NGS after an in-house development and a comprehensive validation study, based on the comparison of PGS results obtained by NGS to those obtained by established aCGH technique (array comparative genomic hybridization), with **more than 100 embryos diagnosed and 100% diagnosis correspondence** by both techniques, only with higher detection power for smaller chromosomal alterations by NGS.

BIOARRAY

PREIMPLANTATION GENETIC DIAGNOSIS
USING
NEXT GENERATION SEQUENCING

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PREIMPLANTATION GENETIC DIAGNOSIS
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Bioarray proudly presents the new **Preimplantation Genetic Diagnosis (PGD) using Next Generation Sequencing (NGS)**. PGD is intended for fertility clinics which want to offer this valuable complementary test for some of their patients, in order to increase implantation chances or to prevent inheritance of a genetic disease, by selecting the most viable embryos obtained in the in vitro fertilization (IVF) procedure.

NGS is currently the most advanced technology for the study of the human genome and Bioarray has pioneered the application of this technique for human reproduction health.

Bioarray offers both kinds of Preimplantation Genetic Testing using NGS:

- Preimplantation Genetic Screening (PGS), consisting of the genetic study of the embryo's genome in order to determine its chromosomal arrangement and in turn, selecting **chromosomally normal (euploid) embryos for transfer, increasing pregnancy success rates in IVF**. NGS allows detection of whole chromosome copy variation, but also detection of small size insertions and deletions (6 Mb approximately), as well as Robertsonian translocations (causing, for example, Down syndrome).
- Preimplantation Genetic Diagnosis (PGD), allowing the detection of a specific disease-causing mutation in the embryos, a powerful tool for those couples resorting to IVF to **prevent a single-gene disorder being passed on to their children** (e.g. cystic fibrosis).

When is DGP recommended?

Main indications for PGD are:

- Advanced maternal age.
- History of recurrent miscarriage.
- Repeated failure in IVF cycles (>2).
- History of pregnancy or children with chromosomal anomalies.
- Men with chromosomal anomalies in sperm.
- Couples carrying a single-factor hereditary disease.

What are the advantages of this new technology?

- PGS using NGS checks **all of the 24 chromosomes with higher resolution, achieving greater diagnostic power**. Previously existing techniques only studied a few specific chromosomes or could only detect full aneuploidy (extra or missing copy of a chromosome).
- PGD using NGS checks for **single-gene disorders when parents carry the condition** (e.g. Cystic Fibrosis, Fragile X syndrome, etc.) and thus at risk of transmitting it to their children.
- When testing for a single-gene disorder (PGD) is requested, Bioarray reports **mutation-free embryos but also chromosomally normal embryos (PGS), all with a single biopsy**, contributing even more to the success of the IVF treatment.
- Using NGS for PGS provides a **more affordable cost** thanks to the NGS high throughput, making it possible for more couples to have access to this test.

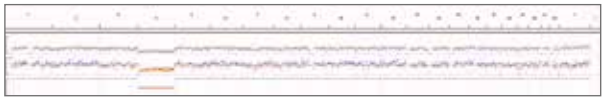
PGS techniques comparison

	FISH	Karyolite	24sure	24sure+	karyomapping	NGS
24 chromosomes		•	•	•	•	•
CNVs* detection (around 6Mb)				•	•	•
Mitochondrial DNA						•
Single-gene disorders					•	•
Price	$	$	$$	$$$	$$$	$
Technology	FISH	Luminex	Microarray CGH	Microarray CGH	Microarray SNPs	Massive sequencing

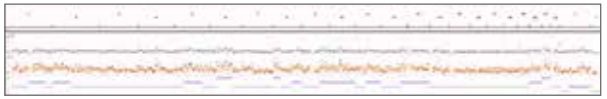
*copy number variation

Result graphs

Normal Male (46 XY)



Abnormal female (45 XX) - chromosome 4 monosomy



Caotic - multiple abnormalities

Graphs show levels of the NGS sequence readouts throughout the 24 chromosomes, thus revealing existing aneuploidies in case of increased or decreased readout level.

NGS: Present and future of genetic analysis

NGS massive parallel sequencing technologies have been a major breakthrough in the field of genetic research, since they allow sequencing of great amounts of DNA at a reasonable cost.

NGS methodology applied for PGD offers the possibility of obtaining a large number of sequence readouts, uniformly distributed throughout the embryo's genome, including the mitochondrial DNA, in order to detect the possible occurrence of chromosomal structural abnormalities, as well as single-gene disorders.

Bioarray's NGS is backed by a highly experienced bioinformatics team, an essential area of genetic analysis and a key aspect in PGD too when for example dealing with the Allele Dropout phenomenon.