# Preimplantation genetic diagnosis in asymptomatic carriers

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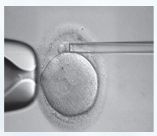
## Introduction

Preimplantation genetic diagnosis (PGD) is defined as the in vitro testing for Mendelian, chromosomal and mitochondrial defects in preimplantation stages embryos or oocytes and an alternative for pre-natal diagnosis and selective termination of pregnancy in couples with a high risk of affected offspring.

There are some detractor groups against the application of PGD in general, although some indications are specially ethically difficult, such as non-disclosure PGD for late-onset diseases: patients do not wish to know their carrier status but want to have disease-free offspring.

## Material and methods

1. **Assisted Reproductive Technology** (**a.** IVF; **b.** ICSI)
2. **Genetic material obtaining** (**a.** Biopsy of polar-bodies; **b.** Biopsy of blastomeres; **c.** Biopsy of blastocyst cells)
3. **Analysis of cells obtained** (**a.** Molecular analysis; **b.** Cytogenetic analysis)

1.a. 
1.b. 
2.a. 
2.b. 
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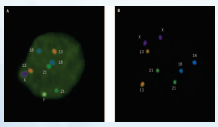
3.b. 

## Ethical issues

**1. Non-disclosure PGD (asymptomatic carrier parents)**

✓ Parents do not want to know their carrier status but do want to have disease-free offspring

✖ Puts practitioners in a difficult ethical position:
- Undertake PGD cycles even when the results of previous cycles preclude the individual as a carrier
- Having to do mock-transfers if no embryos are available

→ The ESHRE ethics task force encourage exclusion testing (the mutation itself is not analysed)

**2. Late-onset diseases and cancer predisposition**

- **Late-onset diseases (such as Huntington's disease and Alzheimer's disease)**

✓ Usually complete penetrance and incurable

✓ Burden imposed by the eventual fate in late-onset diseases

✖ Offspring have a normal life before the onset of the disease

- **Cancer predisposition**

✓ Severe disease

✓ Burden on the quality of life imposed by far-reaching preventive measures in cancer predisposition syndromes

✖ Genetic cancer predisposition doesn't ensure the development of the disease

**3. Carrier embryos**

✓ Pathology disappears of the family

✓ Offspring won't have to use techniques such as PGD to avoid the disease in their own offspring

✖ Negative selection of healthy carrier embryos

✖ If there are only carrier embryos available, parents will have to decide to transfer them or not

## Legislation

According to the Spanish Law 14/2006, Preimplantation Genetic Diagnosis with purposes other than the detection of early-onset severe diseases or alterations that may compromise the viability of the embryo requires the approval of the competent sanitary authority after a favorable report from the National Human Assisted Reproduction Commission, which must evaluate the clinical, therapeutic and social features in each case.

## Conclusions

- Preimplantation genetic diagnosis is a useful tool for preventing hereditary genetic diseases and increases the probability of a successful pregnancy in assisted reproduction techniques.

- Nowadays, PGD enables diagnosis of a broad range of genetic diseases but there are many not diagnosable diseases, its efficiency is still limited and it presents some detractor sectors that critic its application.

→ However, some experts call *PGD the medicine of the future.*

## Bibliography and References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**Figures:**

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