Gynecology and Reproductive Endocrinology

Preimplantation genetic testing for HLA matching: a systematic summary of 728 embryos analyses

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Abstract

Preimplantation genetic testing (PGT) requires the use of assisted reproductive technology (ART) to create several embryos, followed by biopsy of embryonic cells for genetic testing and transfer of selected embryos to the womb to establish a pregnancy. PGT-M refers to testing for nuclear DNA mutations causing monogenic disorders, with an autosomal dominant, autosomal recessive or X-linked transmission pattern, but also mitochondrial DNA mutations. It also refers to exclusion testing and to HLA typing with or without concurrent testing for a monogenic disorder. HLA typing of ART-created embryos was first reported in 2001. The aim is to establish a pregnancy that is HLA-compatible with an affected sibling who requires haematopoietic stem cell transplantation. HLA-typing can be performed with or without PGD for the exclusion of a single-gene disorder.

The principle of the methods is based on haplotyping (i.e. determination of the group of alleles within a genetic segment on a single chromosome being inherited together). Therefore, genetic markers located close to the gene of interest are genotyped in DNA samples of the couple and relevant family members with known genetic status during preclinical work-up. Genetic markers that are informative, flank the locus of interest and allow discrimination of the parental haplotypes, are selected for use in the clinical test.

The HLA typing was performed by using short tandem repeats (STRs) as microsatellite markers present in the HLA locus (Fig.).

We analyzed trophectoderm biopsy from 728 embryos and a total of 123 embryos (16.89%) were HLA compatible. PGT-M was tested for 8 different monogenic disorders in 82% (597/728) of all embryos, such as thalassaemia and sickle cell anaemia. For the remining 18% of embryos , the indication for HLA -matching was for healthy embryos could matched with the affected child so that cord blood from the future newborn can be used to transplant the sibling for other medical reasons.

Biography

I graduated in medicine in 1999, and start my residency in 2001 in medical genetics (São Paulo University / SP) which was done tree years later. In additional, in 2002, I undertook a fellow at University of Toronto doing molecular cytogenetics (SKY and CGH) on samples from my patients with chromosome rearrangement. I have my title of specialist in clinical genetics by the Brazilian Society of Clinical Genetics, and also had my PhD title working on molecular biology with Preimplantation Genetics Testing (PGT). Now, I am a head of genetics department in private lab and involved in projects to develop new approaches to detect genetic diseases on non invasive prenatal diagnosis (NIPT) and PGT. I have more that 20 papers published on the international scientific medical journal which all are in the pubmed, over that 50 poster presentations in Brazilian and international lectures of medical genetics. In addition, I am a speaker at the main medical congresses in Brazil.

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