



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.).

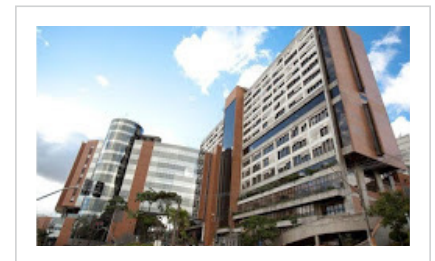
We analyzed trophectoderm biopsy from 728 embryos and a total of 123 embryos (16.89%) were HLA compatible. PGT-M was tested for 8 different monogenic disorders in 82% (597/728) of all embryos, such as thalassaemia and sickle cell anaemia. For the remaining 18% of embryos, the indication for HLA -matching was for healthy embryos could be 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

CiroDreschMartinhago graduated in medicine in 1999, and started his residency in 2001 in medical genetics (Sã o Paulo University / SP) which was done three years later. In addition, in 2002, he undertook a fellow at University of Toronto doing molecular cytogenetics (SKY and CGH) on samples from his patients with chromosome rearrangement.

Publications

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