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کنگره بین المللی
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Medical Genetics Laboratory of Dr.Zeinali

Molecular PGD in Iran: reporting experience on more than 500 blastomeres

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Background

Preimplantation genetic diagnosis (PGD) has been developed to detect genetic disorders before pregnancy. In this method diagnoses performed on blastomeres biopsied from 8-cell stage embryos created by in vitro fertilization method (IVF). Molecular PGD is a PCR-based approach which is used in Dr. Zeinali's Medical Genetics Lab. This study was approved by research committee of Kawsar Human Genetics Research Center.

Method

More than 100 unrelated Iranian families referred to our laboratory. After genetic consolation, peripheral blood samples were collected in tubes containing EDTA and genomic DNA was extracted using salting out method. Mutation detection on samples was carried out using direct sequencing method. For each case fragment analysis and haplotype mapping performed to trace defective alleles in family using multiplex short tandem repeats (STRs). Informative STR markers and selected mutation are checked on each blastomere using nested PCR method.

Result

We have performed PGDs for 76 cases (527 blastomeric) since 2009. 10 families have proceeded to pregnancies and child birth and 2 of them are still pregnant. Other cases were either nontransferable or their implantations did not end up to successful pregnancies regards reduced successful rate of IVF.

Table illustrating cases referred for PGD

	Sex selection	Aneuploidy detection	HLA typing	-
Beta thalassemia	7.59%	5.88%	14.04%	12.14%
Hemophilia A	6.07%	5.31%	-	-
Hemophilia B	3.03%	-	-	-
PKU Type I (PAH)	4.74%	-	-	-
PKU Type II (PTS)	1.70%	-	-	-
DMD	3.03%	1.7%	-	-
Fanconi anemia	-	-	8.72%	-
Deafness	1.51%	-	-	1.51%
EB	-	-	3.59%	-
FHL4	-	-	1.51%	-
CF	-	1.32%	-	-
-	7.40%	-	-	-
Aneuploidy detection	9.10%	-	-	-

Conclusion

PGD is regarded as a powerful diagnostic tool, for carrier couples who desire a healthy child and wish to avoid medical abortion. Results obtained from haplotype mapping in parallel with direct mutation detection make this method a reliable technique with 99.9% accuracy.

Keywords

PGD, Blastomere, Nested PCR, STR, Haplotype mapping, Iran.