17TH Annual International Conference on Preinplantation Genetics
P-37
Bangkok, Thailand 3-6 May, 2018

Aspect as carrier diagnosis of peimplantation genetic testing for monogenic of X-linked recessive disorder

Ammae M[1)], Nakano T[1)], Matsumoto Y[1)], Yamauchi H[1)], Ota S[1)], Nakaoka Y[1)], Morimoto Y[2)]

1) Sunkaky Medical Corporation IVF Namba Clinic
1-17-28 Minamihorie, Nishi-ku, Osaka Japan, Zip 550-0015
TEL: +81-(0)6-6534-8824
FAX: +81-(0)6-6534-8876
E-mail: ammae458@ivfjapan.com
2) Sunkaky Medical Corporation HORAC Grand Front Osaka Clinic
15th floor, Grand Front Osaka Tower B 3-1, Ofuka-cho, Kita-ku, Osaka, Japan, Zip 530-0011
TEL: +81-(0)6-6377-8824
FAX: +81-(0)6-6377-8834

Introduction : The aim of this report is to explore the situation surrounding the preimplantation genetic testing for monogenic (PGT-M) in Japan. PGT-M in Japan started in 2004 when Japan Society of Obstetrics and Gynecology approved clinical research for Duchenne muscular dystrophy (DMD). In Japan, only the molecular genetic testing, which directly analyze the target gene, is approved as diagnosis method. But cases of PGT-M are much fewer than prenatal diagnosis, and the guideline for PGT-M has not developed yet. Accordingly, guideline for prenatal diagnosis is being used for PGT-M. In prenatal diagnosis, by common law, genetic testing would only undergo after the result of sex discernment is revealed to be male. If it is female, whether be a carrier or be healthy cannot be distinguished. By contrast, PGT-M inevitably become carrier diagnosis without sex discernment, because it must be directly analyses method in Japan. However, client has no option to select wheter to transfer or not the carrier blastocyst in PGT-M.

Material & methods : Since April 2016, 10 couples wished to take X-Linked PGT-M including 5 cases of DMD, 3 cases of Adrenoleukodystrophy and 2 others. By findings gained through the genetic counseling, this report will consider the couple's needs for PGT-M.

Results : All the couples had gotten the explanation about the prenatal diagnosis. They all stated that since there was no option to abort a carrier while they were pregnant, so the carrier diagnosis was meaningless in prenatal diagnosis. Even if the male was pathogenic variant, they wanted to avoid

abortion, which was the most important reason that they wanted to undergo PGT-M.
Furthermore, some also stated that they were suffering the conflict about the carrier diagnosis in PGT-M. The carrier would be healthy, and would not onset of X-linked recessive disorder. However, their conflict was whether they would transfer the carrier blastocyst after knowing they must deliver a carrier just like themselves in the future. They felt painful making their child suffer the same way as they had done.

Conclusion : Genetic counselors support clients to make their own decision. We, as genetic expert, shall not regard PGT-M same as prenatal diagnosis any more. It is expected developing guideline specified for PGT-M in Japan.