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Ammae M, Nakano T, Yamauchi H, Kadogami D, Konishi H, Nakaoka Y
Morimoto Y

Considerations on the handling on carrier embryos of X-linked disorders in PGT-M

The Japan Society of Obstetrics and Gynecology (JSOG) has indicated in the ethical guidelines that both the direct method, which examines pathogenic variants, and the indirect method, which performs haplotype analysis, should be used for the diagnosis in preimplantation genetic testing for monogenic (PGT-M). By testing with these two methods, X-linked disorders can be diagnosed not only in normal and abnormal embryos, but also in carrier embryos. On the other hand, the JSOG recommends that normal and carrier embryos should be treated as unaffected embryos. In this study, we retrospectively examined how couples perceive carrier embryos.
The subjects were 38 couples with X-linked disorders who visited our clinic seeking PGT-M. Of the 38 couples, 26 applied for PGT-M, 23 were approved, and 18 underwent PGT-M. All couples expressed opposition to treating carrier embryos in the same way as normal embryos and wished to avoid transferring them. Following PGT-M, the final four couples had only carrier embryos available for transfer. Of these, three couples proceeded with the transfer, resulting in one couple having a live birth.
Although carrier embryos do not develop severe hereditary diseases that indicate PGT-M, there is a risk of giving birth to an affected child in future generations. The couple objects to treating carrier embryos in the same way as normal embryos. The majority of couples expressed a desire for autonomy in deciding how to handle carrier embryos. It is desirable to disclose the PGT-M results and allow couples to make informed choices.