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the couples' choices on preimplantation genetic testing for monogenic after genetic counseling in Japan

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Introduction : In Japan, preimplantation genetic testing for monogenic (PGT-M) can only be done under Japan Society of Obstetrics and Gynecology (JSOG) approval facility. Inside the approval facility, case has to get an approval from JSOG. Due to such strict condition, there are only 5 facilities that have experienced the PGT-M. Even to the genetic diseases that are subjects of PGT-M among other countries, it is not so in Japan and only limited to severe genetic diseases with onset at childhood. In this study, we will consider the issues of PGT-M in Japan from our genetic counseling and their subsequent decision by couples who wanted PGT-M at our clinic.

Material & methods : The subject is 31 couples who made reservation of consultation with our clinic for the purpose of PGT-M from August 2014. In order to obtain approval of PGT-M by JSOG, the couple need to receive genetic counseling not only at the PGT-M implementation facility but also at two different facilities.

Results : 21 couples out of 30 couples who received genetic counseling at our clinic wished to apply PGT-M to JSOG.
A couple with a child with an Alpha-thalassemia X-linked mental retardation syndrome failed to undergo consultation for genetic counseling at our clinic because the condition of the child worsened.
After genetic counseling at our clinic, seven couples did not want PGT-M.
One couple with a child with congenital glycosylation disorder of glycosylation (type Ik) gave up PGT-M because it was difficult to attend our clinic with treatment of the affected child since it took more than 3 hours to our clinic. Four couples abandoned PGT-M because it takes a long time to approve.

Also, in a couple with Myotonic dystrophy 1 male patient, the fact that they are not suited for PGT-M was not easily accepted and they had to be re-consulted by other facilities before they could accept the fact. The couple of congenital myopathy central core disease was selected for PGT-M because of adult onset disease.
In other two couples, one couple applied for PGT-M at different facility, and the other has not yet been able to make the decision after genetic counseling.

Conclusions : Although there may be couples who want to take PGT-M, there are not enough facilities that have experienced the PGT-M in Japan. In addition, indication of PGT-M in Japan is severely limited to diseases in which symptoms that strongly disdain daily life are developed and the survival is dangerous before reaching adults. In the future, based on overseas adaptation criteria, Japan's adaptation criteria should be relaxed