Title: Analysis of 19 cases of autosomal recessive genetic disorders consulted for PGT-M

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[Purpose] We analyzed cases of autosomal recessive (AR) genetic disorders in patients who visited our clinic seeking preimplantation genetic testing for monogenic disorders (PGT-M).

[Methods] Between April 2014 and April 2023, 19 couples with 17 different AR diseases consulted our clinic. After genetic counseling, five couples did not wish to apply to the Japan Society of Obstetrics and Gynecology (JSOG). Out of the 12 couples who applied, nine were approved, two were rejected, and one application is currently under review by JSOG. The reason for one rejection was that congenital nephrotic syndrome, having treatment options like kidney transplant, did not meet the severity criteria. The other case was not approved because the gene variant was considered a variant of unknown significance.

[Results] PGT-M was actually performed in five cases. The average age of the women was 37.7 years. Over nine oocyte retrieval cycles, the average number of oocytes retrieved was 11.1, and the average number of biopsied blastocysts was 4.9. Of the 44 blastocysts analyzed in total, 32 (72.7%) were unaffected, 9 (20.5%) were affected, and 3 (6.8%) were undetermined. We performed embryo transfers over 10 cycles, and the pregnancy rate per embryo transfer was 60%. The miscarriage rate was 10%, and live birth was achieved in all five cases (two of which are ongoing pregnancies).

[Conclusion] The results of PGT-M treatment for AR diseases were promising. PGT-M for AR diseases, including very rare disorders, requires closer collaboration with geneticists and experts in the respective diseases.