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Conflict of Interest Disclosure
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Title:

Evaluation of genetic diagnoses in couples requesting preimplantation genetic testing for monogenic disorders (PGT-M)

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Objective:
For preimplantation genetic testing for monogenic disorders (PGT-M) to be approved by the Japan Society of Obstetrics and Gynecology, the identified variant must be classified as pathogenic. Certain variants associated with rare diseases or lacking prior documentation present significant challenges in the assessment of pathogenicity. In this study, we evaluated genetic diagnoses in couples who requested PGT-M based on the outcome of their PGT-M application.
Materials and Methods:
We retrospectively reviewed genetic testing reports of couples who visited our clinic for PGT-M consultation between 2016 and May 2025. We analyzed the type of institution conducting the genetic testing, the interpretation of the identified variants, and the outcomes of the PGT-M applications.
Results:
Of the 109 couples evaluated, 34 underwent genetic analysis conducted by research institutions, including universities. Among the cases submitted for PGT-M, two variants were classified as of uncertain significance, and one was associated with a susceptibility gene. As none of these variants were deemed pathogenic, approval for PGT-M was ultimately not granted.
Conclusion:
Some variants were not assessed as pathogenic, resulting in ineligibility for PGT-M. Variants diagnosed in a research context may later be used for clinical purposes, including PGT-M and prenatal testing. Thus, adequate genetic counseling, with careful interpretation of variants, is

essential not only at facilities performing PGT-M but also at the institutions conducting the initial genetic analysis.