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Case report: Two cases of oocyte maturation disorder due to specific pathogenic gene

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Introduction: In cases of oocyte maturation disorders, repeated egg retrievals may result in the failure to obtain fertilized eggs. Infertility-related genes through whole-exome sequencing were identified in two cases of oocyte maturation disorder at our facility. Therefore, we report the details of their clinical courses and process of genetic analyses.

Methods: After providing information on two couples who had undergone egg retrievals without obtaining fertilized egg, genetic counseling was conducted, and genetic analysis was performed with informed consent. The present study was approved by the Ethics Review Committee of Fujita Health University.

Results: Case 1: A 27-year-old woman with no history of pregnancy or childbirth. AMH: 3.43 ng/mL. She underwent 12 cycles of egg retrieval, with a total of 127 eggs collected and only 23 eggs matured (maturation rate of 18.1%). Conventional IVF, ICSI, and one day old were attempted, but no fertilized eggs were obtained. Genetic analysis revealed a heterozygous variant in *TUBB8*: NM_177987.3:c.1164G>A (p.Met388Ile). Case 2: A 32-year-old woman with no history of pregnancy or childbirth. AMH: 5.59 ng/mL. She underwent 9 cycles of egg retrieval at a previous clinic, with a total of 82 eggs retrieved and only 5 eggs matured (maturation rate of 6.1%), but no fertilized eggs were obtained. At our facility, 1 cycle of in vitro maturation (IVM) was performed, with 9 eggs retrieved and 0 egg matured in vitro. Genetic analysis revealed a compound heterozygous variant in *PATL2*: NM_001387263.1:c.1225-2A>G and NM_001387263.1:c.1181C>G (p.Thr394Ser).

Conclusions: *TUBB8* is a primate-specific gene, and its pathogenic variants have been reported to cause oocyte maturation disorders and embryonic developmental defects. Additionally, *PATL2*-deficient mice exhibit impaired oocyte maturation, and in humans, *PATL2* variants have also been reported to cause oocyte maturation disorders. In the present study, infertility-related genes were identified in two cases of oocyte maturation disorder,

leading to the decision to avoid further egg retrieval. Since egg retrieval increases the physical, emotional, and financial burden on the patient, identifying the cause of infertility is extremely important. Clarifying the patient's genetic background may lead to termination of infertility treatment or affect fertility of their blood relatives, therefore careful and sufficient genetic counseling are warranted before testing.