P01.049.A - A case report of a rare nonsense *ZP1* variant in a patient with oocyte maturation defect

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Disclosures

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Abstract

Introduction: Oocyte maturation defect (OOMD) is a rare condition causing female infertility that can be diagnosed during assisted reproduction techniques (ART). OOMD related genes are *ZP1*, *ZP2*, *ZP3*, *PANX1*, *PATL2*, *TUBB8*, *WEE2* (OMIM, 2020). We report a case of a 31-year-old woman who had four ART failures diagnosed as empty follicle syndrome and OOMD. She has short stature (-3 SD), bilateral limited extension-flexion on elbows.
Materials and Methods: Chromosome analysis and fluorescence in-situ hybridization (FISH) using X chromosome centromeric and SHOX-probe on interphase nuclei of lymphocytes and mucosal cells was investigated. Whole-exome sequencing (WES) performed via the Illumina platform. Confirmation and familial segregation analysis were performed by Sanger sequencing. **Results:** Karyotyping and FISH resulted in normal, possible mosaicism was excluded. WES analysis revealed a known, rare, pathogenic homozygous variant in exon 3 (c.628C>T; p.Q210*) of *ZP1* gene, and her parents being first degree cousins were carriers for this variant.
Conclusions: *ZP1* with autosomal recessive inheritance is related to OOMD-1 (MIM_615774). Zona pellucida (ZP) is a glycoprotein structure surrounding oocytes and is essential for oocyte development. ZP contains four types of receptor proteins (ZP1-4). Our variant in *ZP1* is nonsense, premature stop codon causes to truncate ZP1 receptor proteins. This is the first homozygous occurrence of this variant associated with OOMD. WES findings were also analyzed for known genes related to short stature and no pathogenic variant has been observed. WES is a valuable method to identify the genetic origin in complex, multigenic conditions like in infertility.Istanbul University Project-Number:TSA-2018-32135