Macrozoospermia associated with mutations of AURKC gene: First case report in Latin America and literature review

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Abstract

A Chilean 35-year-old male patient with a history of primary infertility made an appointment at the Unit of Reproductive Medicine at Clinica Las Condes, Santiago, Chile. Multiple semen analyses revealed abnormal sperm morphology as the most prevalent finding. Multiflagellated and macrocephalic spermatozoa were observed and indicated a possible macrozoospermic phenotype. The constant presence of abnormal sperm morphology led the scope of the study to include Aurora Kinase C (AURKC) gene sequencing. The patient was diagnosed with a homozygous mutation of this gene. The mutation was detected in exon 6, type c.744C>G(+/+) (P.Y248*) variant. As previously described in the Human Gene Mutation Database (HGMD), this pathogenic variant is associated with macrozoospermia. Although this mutation is not the most frequently observed, it is the first of its kind reported in Latin America.

Palabras clave

Palabras clave de autor: Mate infertility; Sperrniorarn; Sperm morphology; Tratozoospermi; Macrozoospermia; Aurora Kinase C gene
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