



MyFertility DNA Test Report (MALE)

You have one genetic variant that is associated with fertilisation failure at In Vitro Fertilisation.

Fertilisation Probability Score (FPS)	1% chance of IVF success
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Gene	Genetic variant	Classification	Zygoty
CATSPER EPSILON	c.2393_2398del (rs761237686)	Highly deleterious	Homozygous

Interpretation and further consideration

FPS is a Fertility Genomics proprietary algorithm that calculates the probability of IVF success by incorporating both the effect and rarity of your mutation(s).

You are homozygous (carry two mutated copies) for a 6 base-pair deletion in exon 18 of the CatSper Epsilon subunit gene. The protein encoded by this gene is part of the mature calcium ion channel in sperm. This sperm-specific ion channel is essential for in vivo and in vitro fertilisation. A published case (Brown *et al.*, 2018) of a man with this mutation reported that mutant sperm fail to fertilise at IVF.

Fertilisation was achieved using Intracytoplasmic Sperm Injection (ICSI).

This report is not intended to be diagnostic, definitive or replace the advice of your physician. Assisted conception treatment always carries a risk of failure due to unknown and unpredictable biological and technical failures.

Technical Information

Exome capture is achieved using Agilent SureSelect Human All ExonV6. The Agilent SureSelect Human All ExonV6 cover approximately 99% of RefSeq database targets. We guarantee that $\geq 80\%$ of bases have a sequencing quality score $\geq Q30$. See Limitations of Service in our Terms of Service for more information regarding limitations of the test.

References

Brown *et al.*, 2018. Homozygous in-frame deletion in CATSPERE in a man producing spermatozoa with loss of CatSper function and compromised fertilizing capacity. Human Reproduction. 33 (10). 1812-16. PMID [30239785](https://pubmed.ncbi.nlm.nih.gov/30239785/).