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Sperm Mitochondrial Mutations as a Cause of Low Sperm Motility

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We report the unique case of a 28-year-old man who, in spite of having a varicocele and a sperm concentration of 5 million/mL, of which 10% were motile and 20% had normal forms (oligoasthenoteratozoospermia [OAT]), was fertile. This was confirmed by paternity testing using 16 autosomal and 6 Y-chromosomal short tandem repeat (STR) loci. An analysis of mitochondrial genes that included cytochrome oxidase I (COI), cytochrome oxidase II (COII), adenosine triphosphate synthase6 (ATPase6), ATPase8, transfer ribonucleic acid (tRNA) serine I, tRNA lysine, and NADH dehydrogenase3 (ND3) revealed, for the first time, 9 missense and 27 silent mutations in the sperm's mitochondrial DNA (mtDNA) but not in the DNA from the blood cells. There was a 2-nucleotide deletion in the mitochondrial COII genes, introducing a stop codon, which might be responsible for low sperm motility.

Key words: Infertility, oligoasthenoteratozoospermia, mitochondrial DNA, short tandem repeats

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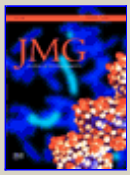

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