

Genetic Morphological Alterations and Sperm Immobility

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The following text is a comment about the article “Homogeneous microscopic abnormalities in sperm morphology and immotility as a cause of male infertility” [1].

The main study question is: What causes morphological alterations to spermatozoa that lead to their immotility? The most frequent causes of total sperm immobility are necrozoospermia and sperm flagella changes. This paper deals with two cases with specific alterations in sperm morphology causing 100% immobility, which are likely of genetic origin: one with thickening of the midpiece, suggesting apoptotic changes and anomalous head-neck attachment causing necrospermia and some headless sperm (patient 1); and another (patient 2) with short, rigid or rudimentary tails, compatible with Primary Ciliary Dyskinesia (PCD) or a related disease as the Dysplasia of the Fibrous Sheath (DFS).

Which was the design of the study (patients/materials and methods)? An observational study for one year on two patients with specific morphological alterations and total sperm immobility was made. There were also seven case-controls included. We carried out a basic and ultrastructural seminal study in both patients. For patient 1 we carried out special stains, DNA fragmentation, FISH, and apoptosis marker study; for patient 2 we performed immunocytochemistry study on sperm flagellum with antibodies against β -tubulin, and a nasal ciliary activity test.

Agreement with Literature

Increases in DNA fragmentation and elevated apoptosis biomarkers

were detected in patient 1, suggesting an apoptotic origin for the abnormal material deposited in the midpiece, which has not previously been described. Other anomalies such as easily decapitated spermatozoa defect (patient 1) and loss of the central pair of microtubules in patient 2 ('9+0' axoneme) have already been reported.

What is the cause of these findings? The familial incidence and the typical phenotype strongly suggest a genetic origin of the syndrome in patient 1; dysplasia of the fibrous sheath is an autosomal recessive genetic disease.

Research Limitations

Patient 1 did not give consent for testicular biopsy to the sperm germ cells study. So it was not possible to determine if the apoptosis detected in this patient's sperm started before or after the ejaculation. The normal nasal ciliary activity excluded PCD in patient 2, making the findings compatible with DFS.

What have we learned with this study? We had find out that homogeneous microscopical abnormalities need specific biochemical and ultrastructural studies in order to determine sperm immotility's origin.

Reference

1. Acevedo D, Ventura J, Aguado C, Carda C, Armengot M, et al. (2014) Homogeneous microscopic abnormalities in sperm morphology and immotility as a cause of male infertility. J Reprod Contracept. 25: 102-118.

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