GENETICS

Mutations underlying azoospermia identified on the X chromosome

Most men with nonobstructive azoospermia do not receive a specific diagnosis. Now, mutations in *TEX11* have been identified that could explain the cause of infertility in some of these men.

Alexander Yatsenko and colleagues used array comparative genomic hybridization to analyse blood samples from 15 men with azoospermia. They found a partial deletion in *TEX11*, which is located on the X chromosome. “The alteration caused meiotic arrest, meaning the precursor cells could not undergo proper meiosis,” explains Yatsenko.

Direct Sanger sequencing of *TEX11* in blood and semen samples from 289 men with azoospermia and 384 control individuals with normal sperm counts identified five novel mutations (three splicing mutations and two missense mutations) in 2.4% of men with azoospermia; no mutations were found in any of the control individuals.

Histological examinations indicated that the mutations were largely detected in patients who had complete meiotic arrest. Mutations were only detected in two patients with azoospermia and mixed testicular atrophy.

“We believe this is the first major genetic cause of male infertility related to a gene on the X chromosome,” says Yatsenko. These findings advance understanding of male infertility and could improve the diagnosis of men with azoospermia and meiotic arrest. “We want to expand our studies and identify other mutations that can lead to abnormal sperm production,” concludes Yatsenko.

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