Sperm Aneuploidy

Chromosomal abnormalities may be somatic cell in origin, in which case they can be detected by a simple blood karyotype analysis. However, most sperm chromosome anomalies arise as a result of errors during meiosis, which cannot be detected by a blood karyotype analysis. These anomalies can only be detected by looking at the sperm chromosomes directly.

Sperm Aneuploidy and Semen Parameters

Approximately 2 to 13% of all sperm are genetically abnormal in normally fertile men. There is evidence that this percentage may be increased in men who are subfertile. Studies have shown a relationship between poor sperm parameters and increased sperm aneuploidy. There is no direct correlation between sperm morphology and aneuploidy, and indeed, sperm aneuploidy can also be found in sperm with normal morphology. However, particular types of morphological defects may be linked to a significant increase in sperm aneuploidy rate, including globozoospermia, amorphous heads, severe tail defects and macrocephalic or multiple head defects.

High Rates of Sperm Aneuploidy and Pregnancy

Studies have shown that sperm with a high rate of aneuploidy have a negative impact on pregnancy rate and are associated with recurrent pregnancy loss.

Measuring sperm aneuploidy

This test uses fluorescent in situ hybridisation (FISH) to label individual chromosomes with specific probes. Hundreds of sperm are assessed from one ejaculate. There are limitations to the test as only 5 probes are currently used routinely for analysis (three of the 22 autosomes: chromosomes 13, 18 and 21, and the sex chromosomes, X and Y), although others are available upon specific request. The results are reported showing incidence of disomy or nullisomy for each of the autosomes and for both sex chromosomes. A sex chromosome ratio is also reported. It usually takes about 14 days to receive the results.

Indications for male patients who may benefit from the test

- unexplained infertility
- multiple failed IVF/ICSI treatment
- recurrent miscarriage in partner
- raised FSH
- oligoasthenoteratozoospermia
- severe teratozoospermia

Causes of sperm aneuploidy

- cigarette smoking
- caffeine
- alcohol
- certain drugs
- chemotherapy and radiotherapy treatment within the last 2 years
- exposure to environmental and occupational pollutants
- exposure to ionising radiation
- advanced age
Treatment

A change in lifestyle may help to reduce these levels in sperm. However, some abnormalities may be irreversible. A recent study indicates that high folate intake may maintain lower aneuploidy rates. There are some studies to show that the use of hyaluronic acid for the selection of genetically healthy sperm for ICSI may be beneficial.

REFERENCES


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