SAT: Sperm Aneuploidy Test
(5 chromosomes: 13, 18, 21, X and Y)

1. **What is the SAT test?**
   The Sperm Aneuploidy Test (SAT) is a diagnostic test to study the genetic etiology of male infertility. In particular, this test evaluates the percentage of spermatozoa with chromosomal abnormalities in a sperm sample. This analysis allows the estimation of the transmission risk of chromosomal abnormalities to the offspring. This test analyzes the chromosomes mostly implicated in spontaneous miscarriages and affected offspring with chromosomal abnormalities (chromosomes 13, 18, 21, X and Y).

2. **Objective. What is the usefulness of the test?**
   At the clinical level, an increase in spermatozoa with chromosomal abnormalities has been associated to a decrease in pregnancy rates and higher miscarriage risk in infertile couples undergoing after ICSI cycles (Rubio et al., 2001; Burrello et al., 2003; Petit et al., 2005; Nicopoullos et al., 2008; Rodrigo et al., 2011; Rodrigo et al., 2013).

   ![](image)

   At the embryo level, different effects have been observed according to the type of sperm chromosomal abnormality. An increase in the percentage of spermatozoa with sex chromosome disomies, results in an increase embryo aneuploides compatible with life (Patau Syndrome, Edwards Syndrome, Down Syndrome, Klinefelter Syndrome, Turner Syndrome and trisomies XXX and XXY). Whereas, an increase in diploid spermatozoa generates an increase in triploid embryos, that mostly miscarried before delivery (Rodrigo et al., 2010).

   At the offspring level, several studies performed in parents of Down, Kinefelter, and Turner syndrome children have shown increases in sperm chromosomal abnormalities associated to the chromosomopathies observed in the children (Eskenazi et al., 2002; Tanget al., 2004; Blanco et al., 1998).

   For these reasons, the Sperm Aneuploidy Test can help in the reproductive genetic counseling of the infertile men to assess the optimal assisted reproduction approach.

3. **Indications. To whom and when?**
   This is a diagnostic test directed to the study of male infertility, indicated to patients with higher sperm aneuploidy risk, that is, men with impaired sperm parameters, mainly oligozoospermia, non-obstructive azoospermia, and severe teratozoospermia.
Another indications, not necessarily associated to impaired sperm parameters are recurrent miscarriage of unknown etiology; couples failing to conceive after several IVF treatments and; couples with a previous pregnancy with chromosomopathy.

4. Advantages. Strengths among other tests?
Compared to classic meiotic studies, this test offers the advantage of assessing the final product of meiosis, the spermatozoon that will directly fertilize the ovum. An average of 2,000 sperm per chromosome is analyzed by two independent observers to obtain clinically relevant results. The percentage of chromosomally abnormal sperm is compared to a control group of fertile normozoospermic donors to assess for statistical differences.

5. Sample. How?
The SAT test can be performed in ejaculated samples and spermatozoa retrieved from the epididymis and testes. Ejaculated samples are obtained after masturbation in a sterile container. In the laboratory the sample is washed with buffered medium and can be stored in the fridge at 4ºC until processing. Spermatozoa retrieved from epididymis and testicle should be processed in a different way. The aspirated sperm or the testicular tissue fragments are placed in a
Petri dish with washing medium and dilacerated with a scalpel, using the standard IVF protocols for testicular sperm extraction.

6. Shipping. How, when, where and who?
The samples should be transported in special container at room temperature with a special packaging to prevent damage during transport. The sample should be addressed to:
IGENOMIX, Laboratorio DGP cromosómico, Parc Científic Universitat de València, C/ Catedrático Agustín Escardino, nº 9, Edificio 3, 46980 Paterna, Valencia (Spain).

7. Sample processing and reporting results. When?
The evaluation of the sperm chromosomal abnormalities takes 2 weeks. Once the analysis is completed, a report is submitted to the patient or clinician.

8. Methodology
The most important steps of the protocol are:
1) Sperm fixation in laboratory slides.
2) Decondensation of the sperm nuclei.
3) Hibridization with fluorescent DNA probes directly targeting the analyzed chromosomes.
4) Fluorescent signal detections, scoring and interpretation under an epifluorescent microscope equipped with specific filters.

9. Limitations
This technique allows the detection of aneuploidy for the limited number of chromosomes included in the test. Seldom in few ejaculated samples or testicular samples, there is no enough spermatozoa for a proper estimation of the risk of aneuploidy.

10. How to start?
Contact with: info@igenomix.com

11. FAQ's
What other chromosomes can be tested?
If a previous chromosomal abnormality is detected in a previous pregnancy, a customized SAT test can be requested and additional FISH probes can be included in the test targeting the implicated chromosomes.

Is sexual abstinence required before collecting the sperm sample for the test?
A previous period of 2-3 days of sexual abstinence is recommended before collecting the sample.
Is there a minimal sperm concentration required to perform a SAT test?
There is not a minimal concentration to request a sperm test, the only requirement is the presence of any sperm, even in a concentration below 1 million sperm/mL and also in samples from the epididymis or testes. Seldom there are not enough sperm to perform the test, and additional samples can be requested.

Is it possible to perform a SAT test in carriers of structural abnormalities?
It is possible to test sperm from carriers of translocations using DNA probes implicated in the chromosome rearrangement.

12. REFERENCES


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