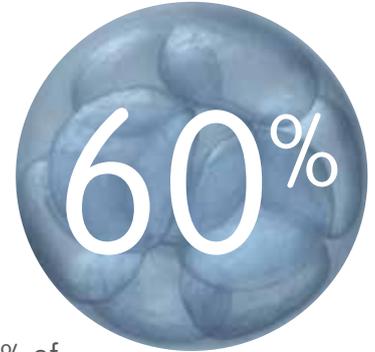


Analysis of sperm chromosomal abnormalities

In couples with severe male factor, there is a higher risk of transmission of sperm chromosomal abnormalities to the offspring.



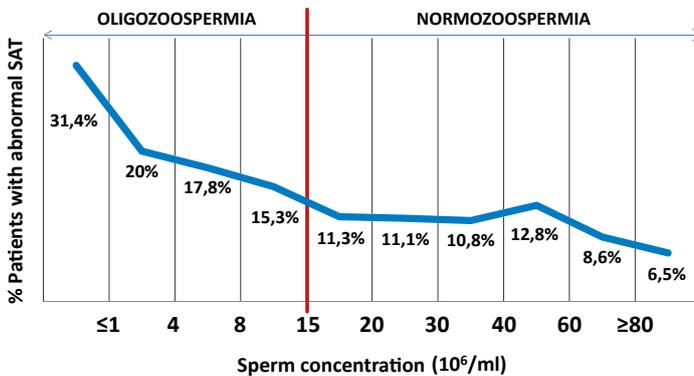
Up to 60% of pregnancies miscarry in couples with an increased incidence of sperm

chromosomal abnormalities

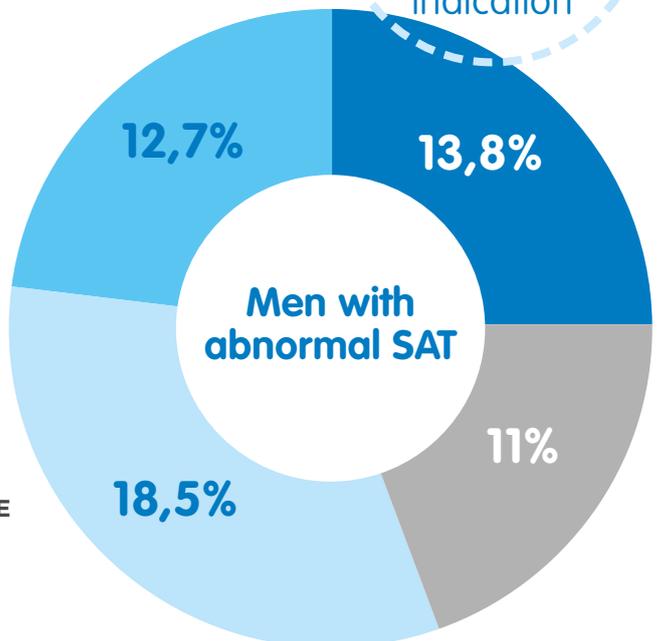
(abnormal SAT)

Indications

1. **Male factor:** the lower sperm concentration the higher incidence of sperm chromosome abnormalities.



Percentage of patients with abnormal SAT for each indication

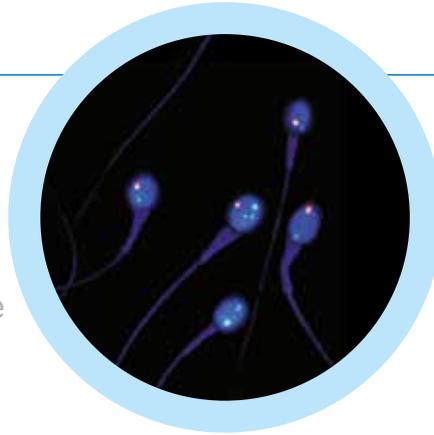


2. **Recurrent miscarriage** of unknown etiology.
3. **Repetitive implantation failure.**
4. **Previous pregnancy** with a chromosomal abnormality.

- MIXED CAUSES** (previous chromosomopathy, chemotherapy, radiotherapy)
- RECURRENT MISCARRIAGE** (≥2 miscarriages)
- REPETITIVE IMPLANTATION FAILURE** (≥2 IVF failures)
- MALE FACTOR** (impaired sperm parameters)

ANALYSIS OF SPERM CHROMOSOMAL ABNORMALITIES

www.igenomix.com



1 What is SAT?

- The Sperm Aneuploidy Test (SAT) is a diagnostic test to study the genetic etiology of male infertility.
- It allows to evaluate the presence of an abnormal number of chromosomes (aneuploidy and diploidy) in the sperm.
- The chromosomes 13, 18, 21, X and Y, mostly implicated in spontaneous miscarriages and affected offspring with chromosomal abnormalities, are analyzed by fluorescence in situ hybridization.

3 Reasons to indicate the sperm chromosomal analysis (SAT).

An increase of sperm chromosomal abnormalities affects to three levels:

EMBRYO LEVEL

- Spermatozoa with sex chromosome abnormalities results in aneuploid embryos.
- Diploid sperm results in triploid embryos.

Rodrigo et al., 2010

PREGNANCY LEVEL

- An altered SAT decreases pregnancy rates after ICSI.
- And increases miscarriage rate. *Rubio et al., 2007*

OFFSPRING LEVEL

It increases the risk of abnormal offspring for the chromosomes affected in the sperm (Down, Klinefelter or Turner's Syndromes)

2 What is the usefulness of SAT?

- The SAT test allows to identify males with low reproductive success chance.
- It is a useful tool to provide a more personalized genetic counseling to the infertile couple previous to perform an in vitro fertilization treatment.
- In couples with an abnormal SAT, it is indicate to perform PGS (Preimplantation Genetic Screening). It allows to select chromosomally normal embryos for transfer, increasing pregnancy rate and decreasing miscarriage risk. *Rodrigo et al., 2014.*

4 How to obtain the sample?

Ejaculated samples are collected in a sterile container and transported to the reference laboratory at room temperature. In the laboratory the sample is washed with buffered medium in a 10 mL conical tube and can be stored in the fridge at 4°C a maximum of 3 days before the shipment to the IGENOMIX lab.

5 How to send the samples?

- The samples should be transported in a sealed conical tube at room temperature with a special packaging to prevent damage during transport.

6 What is the delivery time for results?

- Two weeks would be the maximum time to deliver the results.

two weeks

METHODOLOGY MAIN STEPS OF THE ASSAY



8. Limitations. This technique allows the detection of aneuploidy for the limited number of chromosomes included in the test. In very few ejaculated samples or testicular samples, is there not enough spermatozoa for a proper estimation of the risk of aneuploidy.

Rodrigo L, Peinado V, Mateu E, Remohí J, Pellicer A, Simón C, Gil-Salom M, Rubio C. Impact of different patterns of sperm chromosomal abnormalities on the chromosomal constitution of preimplantation embryos. *Fertil Steril.* 2010 Sep; 94(4):1380-6.

Rubio C, Gil-Salom M, Simón C, Vidal F, Rodrigo L, Mínguez Y, Remohí J, Pellicer A. Incidence of sperm chromosomal abnormalities in a risk population: relationship with sperm quality and ICSI outcome. *Hum Reprod.* 2001 Oct; 16(10):2084-92.

Rodrigo L, Mateu E, Mercader A, Cobo A, Peinado V, Milán M, Al-Asmar N, Campos-Galindo I, García-Herrero S, Mir P, Simón C, Rubio C. New tools for embryo selection: comprehensive chromosome screening by array comparative genomic hybridization. *BioMed Research International.* 2014; In Press.