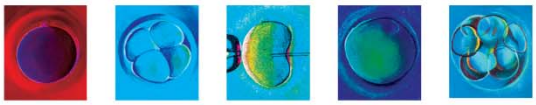
Louis Weckstein, MD
Susan Willman, MD
Mary Hinckley, MD
Deborah Wachs, MD
Carmelo Sgarlata, MD
Evan Rosenbluth, MD
Kristen Ivani, PhD



FERTILITY FACT SHEET

San Ramon / Orinda / San Jose

GENETICS IN MEN WITH SEVERE MALE FACTOR

Criteria for severe male factor:

1. Sperm Count - ≤ 5 million/cc
2. Azoospermia (no sperm)

There are several different genetic conditions associated with male factor infertility:

A. Chromosome Abnormalities:

Approximately 5-10% of men with severe sperm abnormalities have some type of chromosome abnormality, which can be diagnosed by a blood chromosome study called a karyotype. All men who meet the above criteria for severe male factor should consider having this testing done. Men who are found to have a chromosome abnormality have an increased risk of having a live born child with a chromosome abnormality and their sons may also be at increased risk for infertility problems. Some types of prenatal diagnosis, such as amniocentesis or CVS, should be considered if a pregnancy results in this situation to evaluate the chromosome status of the fetus.

B. Y Chromosome Microdeletions:

10 to 20% of men who have severe sperm abnormalities, but normal chromosome number may be missing very tiny areas of their Y chromosome that cannot be picked up on a routine chromosome study. The areas that are missing are called microdeletions and they can be detected using more detailed DNA analysis. There are genes located in these areas that are very important for the production and development of sperm. When these genes are missing ("deleted"), male factor infertility occurs. At this time, it appears that the main implication of having a microdeletion is that it would be passed on to all male children, who would also be infertile. This testing can be considered for men who have a sperm count less than 5 million/cc.

C. Cystic Fibrosis and Congenital Absence of the Vas Deferens (CAVD):

CAVD is the cause of approximately 1-2% of male infertility. This is often the diagnosis in men with azoospermia. CAVD is associated with an increased chance of being a carrier for a gene that causes cystic fibrosis (CF). CF is a recessively inherited condition, meaning both parents must be carriers in order for there to be a risk to have an affected child. CF causes lung damage and digestive problems. Greater than 80% of men with CAVD are carriers for at least one CF mutation.

It is very important that men with azoospermia and/or CAVD and their female partners have CF carrier testing done to most accurately assess their risk of having a child with CF. the male children of men with CAVD may be at increased risk of being affected with CAVD.

Most men with severe sperm abnormalities will need IVF with ICSI (intracytoplasmic sperm injection) for fertilization to occur. Studies have shown that there is a small increased risk of having a child with a chromosome abnormality associated with ICSI. There is thus far no evidence to suggest that ICSI itself causes this risk or that it poses a risk for any other abnormalities. Prenatal diagnosis should be considered for any pregnancy conceived using ICSI.

Men diagnosed with severe male factor infertility may wish to consider having genetic counseling to review this information and have appropriate testing prior to undergoing an ICSI cycle.