SERTOLI CELL-ONLY SYNDROME
Scos, Del Castillo Syndrome, Germ Cell Aplasia

The absence of any developmental stage of sperm cell in the testes.

**Diagnosis**  **Male**

**Related Diagnoses:**
Azoospermia | Klinefelter syndrome

**About Sertoli cell-only syndrome**

Sertoli cell-only syndrome is defined by the complete absence of germ cells in testicular tissues and always results in male infertility. Germ cells give rise to next stages of spermatogenesis which is the process of sperm formation. They are normally attached to Sertoli cells forming together so called seminiferous tubules which form testicular tissue. If there are only Sertoli cells, then no sperm can be produced. The aetiology often remains unknown, yet it is proven that genetic faults takes part in development of this disorder.

**SCOS manifest in two forms, the stage I. and stage II:**

- Stage I. SCOS manifest by complete absence of all spermatogenic cells in the testicles and complete infertility. If there is no morphological alteration of reproductive tract and there is no sperm present in the semen, it is called as non-obstructive azoospermia (NOA).
- On the other hand, men suffering from stage II. SCOS (referred also as focal SCOS) may have at least few germ cells preserved in the testicles, so there is a chance of finding some sperm cells in the testes as the process of spermatogenesis is at least partially preserved.

SCOS is often united with the presence of Klinefelter syndrome. This underlines the relevance of genetic mutations present in SCOS patient. In this particular case, men suffering from Klinefelter syndrome poses more female sex chromosomes than usual. The presence of extra female chromosome interferes with normal development of male body which manifest not only by the look but also by the inability of producing sperm and it usually comes hand in hand with complete sterility of such males.

Another cause of SCOS are the Yq microdeletions which represent another form of genetic alteration of chromosomes. Specifically, the male chromosome called as chromosome Y is affected by missing genes which inflicts malfunction of se development.

Alternative cause to the previous one might be the copy-number variations (CNV). Dogmatic approach states that the genes are present in two copies within the DNA. Quite recently it has been demonstrated that some genes are present in various copies within the DNA. The number of copies varies between individuals. While sometimes it has no effect on the person, other times it can cause serious issues such as SCOS and others.

The diagnosis of Sertoli cell only syndrome is based upon the testicular biopsy. Yet there is a chance that at least partially spermatogenesis can be preserved, most of the testicular tissue is still dysfunctional. Therefore, the diagnosis of specific SCOS stage requires various testicular biopsy to increase the precision.

There is no way to treat the condition itself. Yet there is still way for affected patient to conceive. In most cases, the conception may be achieved through the usage of donated sperm. If the stage II. SCOS is diagnosed, there is still a possibility of retrieving sperm cells directly from the testes. Sperm cells successfully extracted from testicles may be then used in ISCI (intracytoplasmic)-IVF (in vitro fertilization) procedures. Yet surgical extraction of sperm cells from the testicle represents quite challenge, as it is quite difficult to locate functioning
testicular tissue.

**Associated diseases**
- Klinefelter syndrome
- non-obstructive azoospermia

**Complications**
- infertility

**Risk factors**
- genetic predispositions

**Impact on fertility**

The process of sperm formation cannot proceed without germ cells present in the testes. Therefore, males suffering from stage I SCOS are infertile. On the other hand, the stage II SCOS patients may still have chance of conceiving a child. Even though the sperm counts are way too low to conceive a child in “old fashion” way, the assisted reproduction techniques (ART) offer a possible solution.

**Prevention**

There is no way to prevent the development of this disorder. Yet, if parents give birth to child affected by SCOS, it is recommendable to go through genetic testing which can elucidate its origin and help to prevent conception of another affected child.

**Symptoms**

None.

**Therapies**

**Self therapy**

None.

**Conventional medicine**

None.

**Assisted reproduction**

As the SCOS patients most often do not produce any sperm at all, the only possible way to conceive a child is through donated sperm. Sperm donation offers a chance to use verified heathy sperm and there is quite large list of donor from which the patient may choose. Chosen donated sperm is then used to perform intracytoplasmic sperm injection (ICSI) or standard in vitro fertilization (IVF).

ICSI technique involves the injection of single sperm cell with microneedle into an oocyte. Fertilized oocyte is then cultivated in laboratory to a certain stage of development till it is ready to be implemented into a uterus.
IVF technique is based upon simulation of natural process of fertilization. Up to hundreds of sperm cells are used to fertilize an oocyte under laboratory conditions. Fertilized oocyte is then cultivated and implemented into a uterus, when it is ready.

In case of patients suffering from stage II. SCOS, there is way to fertilize an oocyte by the sperm of affected person. To this purpose, the technique called testicular sperm extraction (TESE) is used. A small portion of testicular tissue containing sperm is removed from the testes. Mature sperm cell can be used to perform ICSI. It should be mentioned, that the localization of testicular tissue containing sperm represents quite a challenge and it may fail even after various attempts. The success rates of TESE-ICSI in patients suffering from SCOS are quite low.

Patient whose sperm has been successfully extracted from the testes and used in ISCI-IVF cycle should consider the genetic analysis of early embryo by PGS/PGD (preimplantation genetic screening and diagnosis) technique. This technique allows to study the quality of genetic information of the embryo and may reveal genetic alterations in time.

**Find more about related issues**

### Diagnoses

**Azoospermia**
Complete absence of sperm in the ejaculate of a man.
Learn more at: [www.fertilitypedia.org/therapy/diag/azoospermia](http://www.fertilitypedia.org/therapy/diag/azoospermia)

**Klinefelter syndrome**
The set of symptoms that result from two or more X chromosome in males.
Learn more at: [www.fertilitypedia.org/therapy/diag/klinefelter-syndrome](http://www.fertilitypedia.org/therapy/diag/klinefelter-syndrome)

### Organs

**Testes**
Male gonads which produce both sperm and androgens, such as testosterone, and are active throughout the reproductive lifespan of the male.
Learn more at: [www.fertilitypedia.org/edu/organs/testes](http://www.fertilitypedia.org/edu/organs/testes)

**Uterus**
The uterus is the largest and major organ of the female reproductive tract that is the site of fetal growth and is hormonally responsive
Learn more at: [www.fertilitypedia.org/edu/organs/uterus](http://www.fertilitypedia.org/edu/organs/uterus)

### Reproductive cells

**Oocyte**
A female germ cell involved in reproduction.
Learn more at: [www.fertilitypedia.org/edu/reproductive-cells/oocyte](http://www.fertilitypedia.org/edu/reproductive-cells/oocyte)

**Sertoli cells**
The cell in seminiferous epithelium responsible for nutrition and development of germ (sperm) cells.
Learn more at: [www.fertilitypedia.org/edu/reproductive-cells/sertoli-cells](http://www.fertilitypedia.org/edu/reproductive-cells/sertoli-cells)

**Sperm**
A male reproductive cell which is able to fertilize the counterpart female gamete - the oocyte.
Learn more at: [www.fertilitypedia.org/edu/reproductive-cells/sperm](http://www.fertilitypedia.org/edu/reproductive-cells/sperm)

### Reproductive functions
Spermatogenesis
Process in which spermatozoa are produced from male primordial germ cells in testicles by way of mitosis and meiosis.
Learn more at: www.fertilitypedia.org/edu/reproductive-functions/spermatogenesis

 Symptoms

Infertility
The failure to achieve a clinical pregnancy after 12 months or more of regular unprotected sexual intercourse.
Learn more at: www.fertilitypedia.org/edu/symptoms/infertility

 Therapies

Assisted reproductive technology
The range of medical treatments methods designed to result in pregnancy.
Learn more at: www.fertilitypedia.org/edu/therapies/assisted-reproductive-technology

Egg donation
Process by which a woman donates eggs for purposes of assisted reproduction or biomedical research.
Learn more at: www.fertilitypedia.org/edu/therapies/egg-donation

ICSI
A micromanipulative fertilization technique in which a single sperm is injected directly into an egg.
Learn more at: www.fertilitypedia.org/edu/therapies/icsi

Preimplantation genetic diagnosis
Technology that allows couples with a family history of monogenic disorders, x-linked diseases and chromosomal abnormality have a healthy baby.
Learn more at: www.fertilitypedia.org/edu/therapies/preimplantation-genetic-diagnosis

Preimplantation genetic screening
The term PGS is used to denote procedures that do not look for a specific disease but to identify embryos at risk of de-novo occurring aneuploidies.
Learn more at: www.fertilitypedia.org/edu/therapies/preimplantation-genetic-screening-1

Sperm donation
The procedure in which a man (sperm donor) provides his sperm for fertility treatment.
Learn more at: www.fertilitypedia.org/edu/therapies/sperm-donation

Standard IVF
A process in which an egg is fertilised by sperm outside the body: in vitro. Own or donated gametes may be used.
Learn more at: www.fertilitypedia.org/edu/therapies/standard-ivf

TESE
Removal of a small portion of testicular tissue in order to extract a few viable sperm.
Learn more at: www.fertilitypedia.org/edu/therapies/tese

Sources

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