

Klinefelter's Syndrome and Fertility

11. You can join the KSA

- on-line at www.ksa-uk.net
- via our helpline **0300 111 4748** (option 3)
- by emailing membership@ksa-uk.net

The KSA is run by volunteers and depends on membership fees, fundraising and donations to support its work. Please help us to support others — donate via the website, helpline or contact chair@ksa-uk.net

**PLEASE
DONATE**

Acknowledgements: The KSA would like to thank Dr Kevin McEleny for providing the original copy for this booklet and for his help with the subsequent editing.

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Disclaimer: All information given is general and all patients are different. If you have further questions, please raise them with your medical professionals eg your GP or endocrinologist.

Klinefelter's Syndrome and Fertility



Klinefelter's Syndrome Association

- ☎ Helpline: **0300 111 4748**
- 🌐 Website: www.ksa-uk.net
- 📘 Facebook: **Klinefelter's Syndrome Association**

Klinefelter's Syndrome isn't rare...
but it is rarely diagnosed

eXtra
Klinefelter's
Syndrome
Association

Charity Registration Number 1058319

Klinefelter's Syndrome Association

Klinefelter's Syndrome isn't rare...
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Klinefelter's
Syndrome
Association



The Klinefelter's Syndrome Association is a charity working to support those affected by Klinefelter's Syndrome/XXY.

Klinefelter's Syndrome and Fertility – an overview

1. What is Klinefelter's Syndrome?
2. How does Klinefelter's Syndrome affect fertility?
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8. What other options are available to me?
9. How can I get support and further information?
10. The Klinefelter's Syndrome Association (KSA)
11. Join the KSA

10. The Klinefelter's Syndrome Association (KSA)

The KSA works to:

- raise awareness of KS/XXY and improve diagnostic rates
- reduce the age at which KS/XXY is diagnosed
- encourage and support research

The KSA supports and informs all affected by KS/XXY by the provision of:

- helpline
- website
- Facebook page
- private Facebook groups
- annual conference
- opportunities to participate in research
- activity weekends
- social events
- free membership for medical professionals

Please support the Klinefelter's Syndrome Association (KSA).

Join the KSA to support this important work and to benefit from:

- regular Newsletter
- access to the 'Members only' section of the website
- the KSA Members' Handbook
- access to contact lists
- advance information and reduced cost for annual conference
- subsidised activity weekends
- members only social events

Please turn the page for details of how to join the KSA.

9. How can I get support and further information?

Fertility assessment and treatment can be very stressful. If you would like to see a fertility counsellor to help with this, please ask the team at your clinic to refer you.

If you would find it helpful to make contact with others affected by KS, the Klinefelter's Syndrome Association (KSA) will be able to help (see below).

Sources of help and information

- Klinefelter's Syndrome Association (KSA):
www.ksa-uk.net
- Fertility Network:
www.fertilitynetworkuk.org
- British Fertility Society:
www.britishfertilitysociety.org.uk
- British Infertility Counselling Association:
www.bica.net
- For an overview of Klinefelter's Syndrome see 'Klinefelter's Syndrome', download available at:
www.ksa-uk.net/downloads
- For more detailed information about the effect of XXY on fertility, see 'Male infertility and KS' by Mr. Vinod Nargund; available to KSA members at:
www.ksa-uk.net/downloads/members-handbook



About this booklet

If you have just been diagnosed with Klinefelter's Syndrome (KS) as a cause of infertility, then you may be experiencing a range of emotions and have many questions.

We hope that this leaflet provides some answers for you. It has been written by a specialist in consultation with KS people and contains some basic information about KS and infertility.

At the end of this leaflet you can find sources of more detailed information and support.

1. What is Klinefelter's Syndrome?

KS is a lifelong condition which is present from conception in about 1 in 600 live male births.

Typically, males have one X and one Y sex chromosome (XY), while females have two X sex chromosomes (XX). In KS, males have an extra X chromosome.

The most common KS sex chromosome pattern is XXY and therefore the condition is also known as XXY.

KS is the most common sex chromosome variation affecting humans and is the most frequent chromosomal variation found in men with infertility. It affects the production of testosterone and hence the production of sperm.

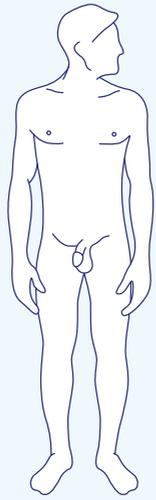
For more details on the symptoms and treatment of KS, please go to the Klinefelter's Syndrome Association (KSA) website www.ksa-uk.net.

Typical Karyotype

46,XY



Higher fertility
Shorter stature
Wider shoulders
Less gynaecomastia
Larger testes

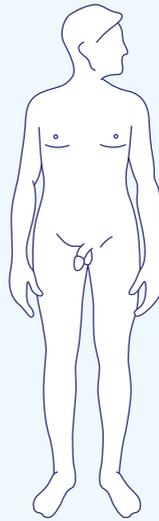


Klinefelter Syndrome

47,XXY



Infertility
May also have:
Tall stature
Narrow shoulders
Gynaecomastia
Small testes



2. How does Klinefelter's Syndrome affect fertility?

Testicles have two roles. One is to make a hormone called testosterone and the other is to make sperm. Both functions are affected by KS, but the degree to which they are affected varies from one person to another.

Men with KS usually have no sperm in their semen. This is because of the way that KS affects the testicles.

The testicles of KS men are at their most active when they go through puberty. Following this they become increasingly less effective at making sperm.

Males with KS usually enter puberty normally, but by mid puberty they may have low levels of testosterone, small testicles and the inability to make sperm.

It is worth bearing in mind, that if sperm are present, the chance of having a baby with one cycle of treatment is roughly 1 in 3. However, this figure can vary from unit to unit and is affected by factors like female age. The fertility team will be able to discuss this with you in more detail.

Access to NHS-funded fertility treatments is limited and unfortunately depends on where you live. As guidelines change frequently, your doctor can advise you on this.



8. What other options are available to me?

Many couples affected by KS choose to use sperm from a donor when trying for a baby. This may involve IVF treatment, but less invasive treatments can also be used. The clinic will explain the process to you, including how to choose a donor.

Sometimes couples ask someone they know, a family member or friend, to be the donor. This is called Known Donation, and for this, the potential donor will need to be assessed by the fertility clinic.

For more information about sperm donor treatment, please visit the National Gamete Donation Trust website (www.ngdt.co.uk).

Some couples choose to adopt or foster. You can find out more about this by contacting your local council or by looking at the following website (www.adoption.uk).

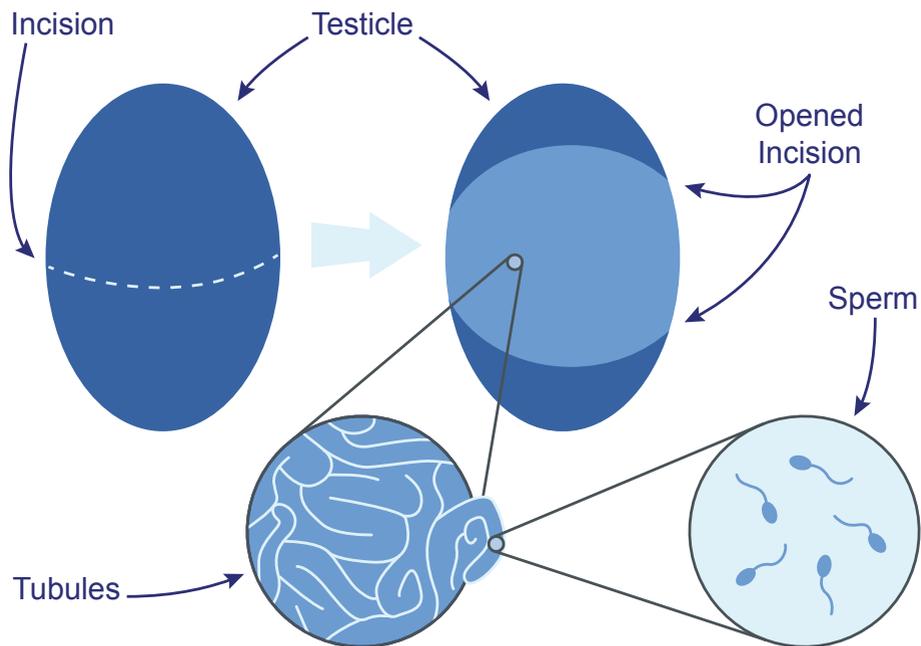
Many couples who can have children choose not to do so. If this increasingly common life choice suits you and your partner, this is as valid a decision as any other.

7. If I don't have sperm in my semen, what are my options?

Sperm can sometimes be extracted directly from the testicles by an operation. There are different sorts of operations used, including ones involving an operating microscope. This operation, illustrated in the diagram below, is called MicroTESE, and some experts feel that it is the best sperm retrieval procedure to perform on men with KS.

The chance of getting sperm with MicroTESE varies from patient to patient. It may be influenced by their age and natural testosterone level, but this isn't certain.

Whilst some experts report that the chance of getting sperm from men with KS can be as high as 50% with MicroTESE, other doctors find their own results considerably lower.

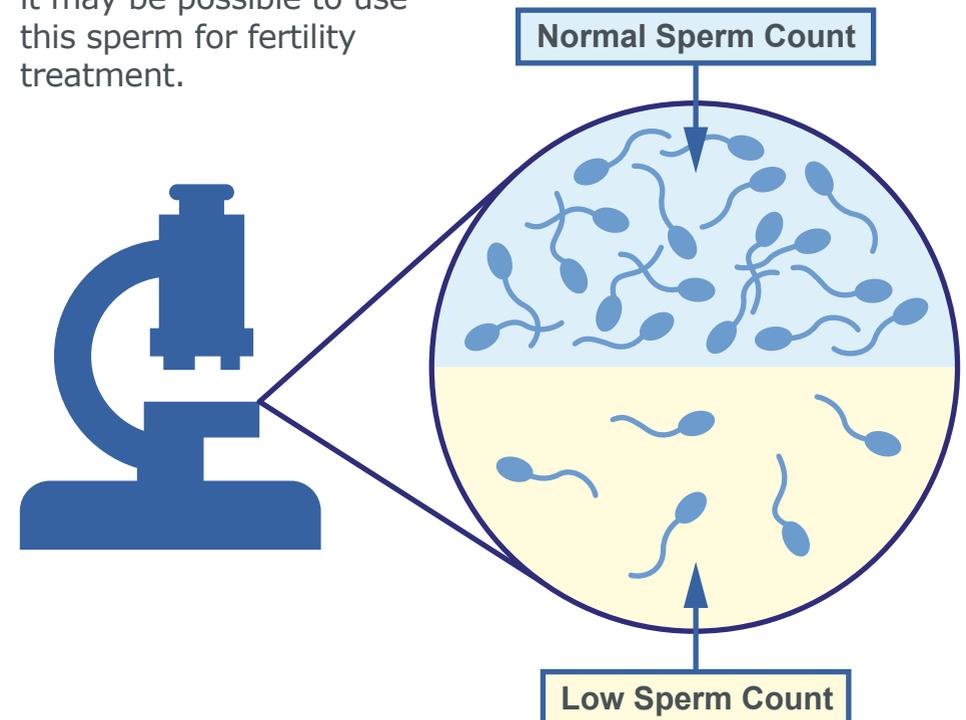


3. Sperm levels and KS

A semen analysis is generally the first test done on men to investigate their fertility. Scientists examine semen samples under a microscope to look for sperm and to assess their concentration, movement and appearance.

Although most KS men don't have any sperm in their semen, some may have small numbers present. If that is the case, it may be possible to use this sperm for fertility treatment.

If you do still have sperm in your semen, your doctor may suggest that you should think about storing them, particularly if you aren't actively trying to have a family at the moment. In that way, your sperm could be used in the future for fertility treatment, even though by then there may no longer be any sperm present in your semen.



4. Testosterone levels and KS

Testosterone is needed for male health. If levels are low, as in the case of many KS people, it can affect mood, energy levels, semen volume and sexual function. However, sometimes low levels of testosterone may not cause obvious symptoms.

Your doctor will check the level by performing a blood test. Blood should be taken in the morning, as afternoon levels aren't accurate.

While many of those with KS have a low testosterone level, some may have a normal level. However, levels generally drop in the 20s or 30s, and most will eventually need testosterone replacement treatment. This is done by using a gel that you apply yourself, or by injection. Your doctor will discuss this with you and keep an eye on your blood test results, to ensure that all is well.

5. Does testosterone treatment affect fertility?

Yes. This is because the concentration of testosterone in the testicles is normally much, much higher than in the blood.

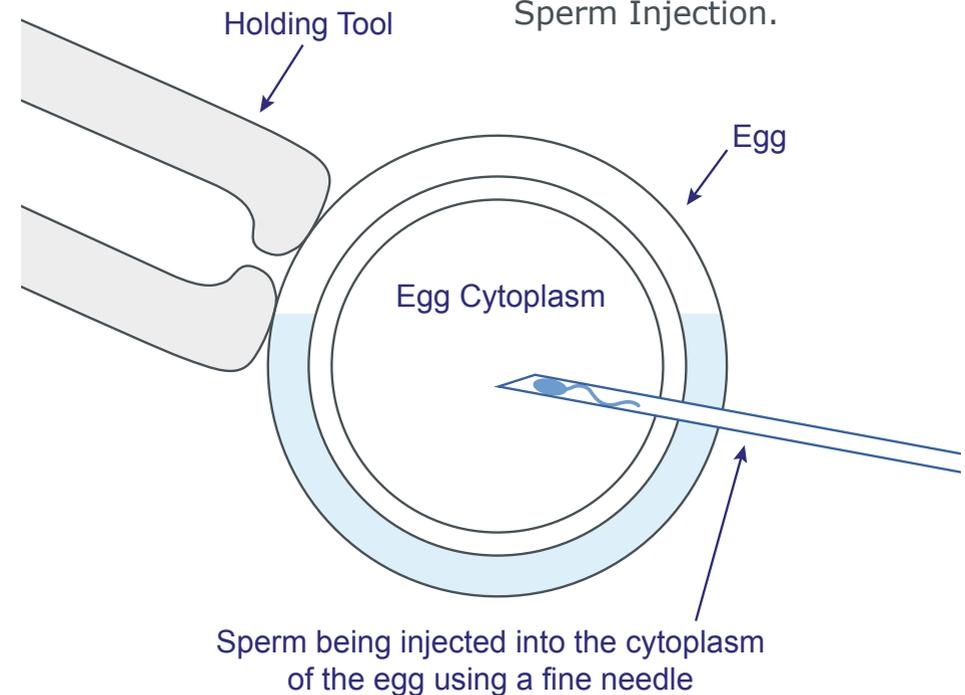
If you are given testosterone treatment, the level in your blood will increase causing the brain to stop stimulating the testicles to make testosterone.

The level in the testicles will then be far lower than it needs to be for sperm to be made normally. This is why, if you are already on testosterone treatment and need to produce a semen sample for fertility testing, your doctor may ask you to stop the testosterone treatment for a few months beforehand. You may be offered alternative medication to help maintain your testosterone levels.

6. If I have sperm in my semen, will I be able to make my partner pregnant?

There have been a couple of cases where this has been reported as happening naturally, but it is extremely unlikely. This is because if sperm are present, the concentration is usually very low.

However, if even very small numbers of sperm are present in the semen, it may be possible to select these sperm and inject them into eggs taken from your partner's body as part of an IVF treatment. This form of IVF treatment is called Intracytoplasmic Sperm Injection.



For more information about fertility treatments please ask your doctor, or visit the Human Fertilisation and Embryology Authority website (www.hfea.gov.uk).