KSA (UK): Frequently Asked Questions

KLINEFELTER'S SYNDROME ASSOCIATION UK

INFORMATION DOWNLOADS

FREQUENTLY ASKED QUESTIONS

Aims of this document

To address the most commonly asked questions from the enquiring public and our members

Note:

The information contained in this paper has been approved by our Medical Advisers but should not be treated as specific advice to individuals. All such information should be checked with your Health Provider. Drug usage in particular is a matter for your Medical Practitioner.

To contact us use any of the following:

Website: http://www.ksa-uk.co.uk/

Mail
National Coordinator
Klinefelter's Syndrome Association
56 Little Yeldham Road
Little Yeldham
Halstead
Essex
CO9 4QT.

Telephone Contact Number: 0845 230 0047
(Calls Charged at Local Rates)

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WHAT PHYSICAL MANIFESTATIONS MAY APPEAR AND HOW SOON?

In a young man with KS, the testicles may not develop normally and therefore the levels of the male hormone, testosterone, will not rise at the onset of puberty as they do in normal young men. This will mean that they will not show the increase in muscle development, and the increase in body hair, normally seen at around the age of 12 or 13 years. The voice will tend not to break, and the young man will generally look less masculine. However, it is quite common for many young men to undergo a normal delay in puberty, as this can vary from between the ages of 12 and 18. The difference in men with KS is that in KS their bones will continue to grow, rather than in constitutional delay where the young man will remain quite short until he eventually goes into puberty. The man with KS will therefore appear to be going into puberty as his growth progresses, but the other features may be absent. The normal rise in testosterone during puberty is required to fuse the bones and stop growth, which will clearly not occur if testosterone is not given at the right time.

As the testicles do not work too well, the rise in the pituitary hormones LH and FSH will try hard to stimulate the glands further, and there maybe an increase in oestrogen rather than testosterone. This may also help delay signs of puberty, and will tend to increase weight gain around the waist. Therefore, some young men with KS will end up slightly pear-shaped due to the high levels of oestrogen.

In the long run, while height can obviously not be changed, weight should be normal, as long as the diet is adequate.

TESTOSTERONE AND TREATMENTS

What exactly are the normal Testosterone levels?

The normal levels of Testosterone in young men after puberty range between 11 and 40 nmol/L. However, there is a wide range in normal levels, and some men are quite happy functioning with levels at 10 or 11 nmol/L without any problem at all. It should also be remembered that some of the testosterone is bound to a protein called SHBG. This means that if the level of SHBG is particularly low, the actual free level of testosterone may still be quite normal even if the total level also appears a little low. Doctors would normally therefore try and look at testosterone in conjunction with the level of SHBG.

As the testicles fail the pituitary tries to work harder and the pituitary hormones FSH and LH will rise. The FSH will rise as a consequence of the lack of sperm formation, and therefore will remain high even with testosterone treatment. The levels of LH will tend to fall with testosterone treatment, but they may still be outside the normal range. This should cause no concern, the critical level to be measured being the testosterone and SHBG (see above).

How is testosterone normally produced?

Testosterone is normally produced by scattered cells within the testicle, which are stimulated by the hormone LH from the pituitary. LH is in turn stimulated by another hormone from a part of the brain called the hypothalamus. In KS the testicles develop poorly and become rather fibrous such that the cells, which make testosterone in the testicle, are destroyed.

Why do KS men not produce it?

In KS the testicles develop poorly and become fibrotic, for reasons which remain unclear. They therefore make subnormal (low or very low) levels of testosterone, although these may still be within the normal range for many years. The levels of testosterone are therefore quite variable in men with KS.

What other blood tests are taken?

Apart from confirming the chromosome disorder in KS, the only blood tests routinely necessary are testosterone, SHBG, LH and FSH.

How do I know if the Testosterone dose is wrong?

There are different types of testosterone treatment, and your doctor will gauge both clinically how well you are doing (how often you shave, the amount of body hair and body muscle etc), as well as taking occasional testosterone and SHBG levels.

What are the side effects of treatment?

Patches, is there a time limit to skin contact per one patch?

The current patches are rather variable, although the amount of testosterone is absorbed really quite well. However, the most common patch, the Andropatch, can often cause skin irritation, and should therefore be applied to a clean, hairless patch of skin, and the patch rotated every day to a fresh site for seven days each week.

Injections?

The most common injections need to be given every one to three weeks according to dose. There is often a very rapid rise in testosterone level after injection, and the levels may become rather low just before the next injection. Your doctor will decide after discussion whether you should have more frequent injections, with all the inconvenience of this, or less frequent injections with the chance that levels of testosterone will be much more variable.

Gel?

There are some good preliminary results with a new testosterone gel, and also with a small patch which fits just behind the upper lip. These preparations should soon be available in the UK.

At what age should any hormone treatment begin?

In general, it would be reasonable to start with small doses of testosterone at or around the age that normal puberty would start. This may vary according to the individual, but somewhere between 14 and 16 years of age seems reasonable, and certainly by 18 years. The doses should probably start low and then gradually build up to the full adult dose over a year or two.

Testosterone treatment should normally be continued throughout life, although the dose may need to be adjusted later in life. There is no evidence for a male menopause, and therefore no age at which testosterone should routinely be stopped.

Does the application of supplementary Testosterone kick start the body's natural production?

If the body is not making testosterone then there is no influence of additional testosterone on the body's own capacity. It will certainly not "kick start" the body's natural production.

Would giving my 18 year old son Testosterone make him aggressive?

Testosterone has a variety of effects on behaviour, in general making men more assertive, and in some situations, rather more aggressive than they might otherwise be. As long as the

levels are kept within the normal range, this is entirely normal. However, patients or their relatives may note an increase in independence and assertiveness and it is always reasonable to start with small doses and build up the dose gradually.

Why does my 27 year old son display verbal aggression after Testosterone injections?

After a testosterone injection the levels may rise to very high levels for a few days in order to stay within the normal range for a full period of three or four weeks. If these early very high levels cause problems, it would be reasonable to use a lower dose more frequently, or transfer to an alternative preparation.

What is the best Testosterone delivery method - patch - implant - injection?

There is no ideal method of testosterone delivery, and new methods are being developed all the time. It is probably best just to be aware that there are a variety of different methods, and these should be customised to the individual patient.

What is the latest news on a gel?

The current news on testosterone gel is that this has been available for some time in the United States but only very recently in the UK. However, there are still currently very few publications on this method, and it may therefore be some time before your doctor is happy to prescribe it.

My 32 year old son's 2 consultants differ in their opinion of the necessity for Testosterone treatment, which should we believe, how can we resolve this

It is extremely difficult to discuss individual cases, and in particular whether or not a given patient should have testosterone treatment in the absence of any further background information. However, there are situations in which individual doctors may come to slightly different opinions. In such circumstances, it is probably best to go along with the doctor with whom one feels most comfortable, and who appears to have the greater experience of this type of condition.

If my son starts testosterone treatment before or at puberty will this prevent breast tissue forming?

Some forms of testosterone can be converted to estrogen, and therefore have the unfortunate side effects of causing breast growth. There is no evidence that starting treatment early will prevent this happening.

BONE DENSITY

Most endocrinologists feel that patients with KS should have bone density scans at some point during their assessment, although if testosterone is started early enough this may not be strictly necessary. Every case needs to be individualised, and there is no automatic requirement for bone density assessment in patients with KS.

How often should I be scanned?

If the bone density is shown to be well within the normal range, there would probably be no reason to repeat the bone density scan, and certainly in not less than five or ten years.

However, if the scan is grossly abnormal, it would be reasonable to repeat this at intervals, although probably not more frequently than once every two years or so.

What grounds would the NHS have for refusal?

The necessity for a bone density scan remains the ultimate decision of the doctor, and his clinical opinion. If he/she believes that a bone density scan is unnecessary, possibly because previous scans were normal or the patient was started early on testosterone replacement, then there cannot be absolute grounds for complaint.

SPERM

Is infertility a certainty?

There are very, very few case reports of patients with KS being able to father children. Although it does occur, it should be assumed that the testicular failure seen in KS is not compatible with fertility.

What about information on freezing sperm for future use (if the testes don't fail immediately)?

The possibility of sperm freezing for future use remains experimental, although it can be carried out in some centres. This should be discussed in some detail with your doctor.

Are KS male's sperm viable (fully formed)?

Even if the sperm are formed, there may be many abnormal forms which swim rather poorly, so even a reasonable count may not necessarily indicate good fertility.

What fertility treatments are available for couples where the man has Klinefelter's Syndrome?

For couples with KS, the main problem is the failure of sperm formation. If there is any evidence of any sperm at all, there are techniques of sperm implantation into the ovum (intra-cytpolasmic sperm insemination, ICSI), which is carried out as part of IVF. If there are no sperm viable at all, then the couple should consider artificial insemination by a donor (AID), or adoption. The availability of AID varies according to the part of the country, as does the willingness of the NHS to fund it.

BREAST TISSUE

Are KS males more vulnerable to lumps or breast cancer?

Because of the testicular failure, the testicle may be pushed to make small amounts of oestrogen, more than that seen in the normal male. This may therefore give rise to an increase in breast tissue. Similarly, testosterone injections can often also be transformed into oestrogen giving rise to excessive breast tissue. For that reason there is a significant, but still very small, increased risk of breast cancer.

Are gynaecomastia operations, viewed by the NHS as necessary, or just cosmetic surgery?

Where there is excessive breast tissue which becomes unsightly, it would be entirely reasonable to be considered for cosmetic repair, including liposuction. However, whether this would be available under the NHS depends on individual consultants and regions. In my

experience, most plastic surgeons would consider this as a necessary operation which could be performed under the NHS.

Can overdeveloped breast tissue be removed by liposuction?

In some situations a small incision can be made just beneath the breast and the excess tissue removed by liposuction. This can be carried out as a day procedure. However, not all plastic surgeons will routinely offer this.

We have been told by a plastic surgeon that surgery would leave a great deal of scarring, what can we do now?

If liposuction is not possible, then a small operation is necessary in which a small incision is made just below the nipple and the excess tissue is removed. This will usually leave a small scar, and possibly some local tethering, but most of the time the scars are relatively trivial. It has to be decided in the individual case whether the improvement in the reduction of the breast tissue is justified in the face of causing a small scar.

If the removal of breast tissue is carried out too early, could there be any further breast development later?

So long as the surgeon is careful to remove all breast tissue, there should not be any regrowth of breast tissue at a later stage. However, it is important that the type of testosterone is then carefully monitored, such that excess levels are not again produced and transformed to estrogens.

OUTLOOK

What is the long-term picture?

In terms of the hormone treatment, there is continuing improvement in the methods of delivery of testosterone, and other than the infertility the male with KS should be assured that he should be rendered entirely normal by means of his testosterone replacement treatment in terms of his bone, muscle, hair growth and behaviour.

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