

Klinefelters Syndrome

Fact Sheet

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Klinefelters syndrome (KS) is a chromosomal abnormality which occurs in approximately one in five hundred men but, because it usually results in infertility, is statistically more frequent in Infertility Clinics, especially those that offer donated sperm. Approximately ten per cent of men with no sperm in their ejaculate (called 'azoospermia') have KS and it is important that chromosomal testing is performed as the Syndrome carries wider implications which will be discussed below.

Explanation

The chromosomal basis is the presence of an extra X chromosome, occurring as a sporadic event. Thus, instead of the usual male configuration of 46XY, these men have 47XXY but, not wanting to confuse the issue, ten per cent of them exhibit mosaicism and have a combination of 46XY and 46XXY. This situation means that some can be offered ICSI (Intra Cytoplasmic Sperm Injection – a more sophisticated form of IVF) using surgically collected sperm and can occasionally be helped to father their own biological children, with normal chromosomes. A review of outcomes in this area is still difficult because of the very small numbers of cases treated in this way, the vast majority achieving parenthood by the use of donor sperm.

Symptoms

Although many men with the Syndrome have a normal appearance and the first discovery of KS is the investigations

for infertility, those with classic KS have gynaecomastia (enlarged breasts) and a tall stature with long legs and a short trunk. There is reduced muscle power and stamina. There are sometimes associated personality and learning difficulties which have been frequently misdiagnosed in earlier life. Depression is common in later life. A single palm crease (Simian crease) is common as is incomplete masculinisation (especially decreased pubic, axillary and facial hair), low libido, a small penis and small firm testes and, of course, the infertility. KS men also have more chronic lung disorders, diabetes, hypothyroidism, varicose veins and breast cancer so they need to have continuing medical surveillance.

Treatment

Hormonal analysis reveals a low testosterone and elevated gonadotrophins and the low testosterone requires long term treatment to improve the masculinisation and to avoid the onset of osteoporosis, address the libido problems, decrease the incidence of depression but it does

nothing to help the sperm problem. Cosmetic surgery (liposuction) is frequently offered for the gynaecomastia with very good effects.

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