KLINEFELTER SYNDROME

Klinefelters syndrome (KS) is not as rare as one may think, it is not life threatening but has life altering consequences. About one in 500 boys are born with an extra X chromosome, this affects learning, behaviour and growth.

This was identified in 1942 by Dr Harry Klinefelter who was working with male patients at a hospital in Boston Massachusetts. This is a genetic condition where a male baby is born with 1 extra X chromosome. This due to an error in meiosis at conception known as meiotic nondisjunction. The condition is also known as XXY. Normal boys are XY while girls are XX. The KS baby develops an abnormal pituitary gland and hypothalamus part of the brain.

Klinefelter syndrome is a random disorder and it is not hereditary. It appears that its occurrence may be linked to the age of either parent. It is not more prevalent in any specific race .This is the most common sex chromosome disorder.

According to research done in Australia only about 20-30% of males with KS are ever diagnosed, partly due to the hesitancy of men to seek medical attention and the somewhat non-specific nature of the symptoms. Often men only find out they are KS when they present with fertility issues. They are almost always sterile.

Unfortunately there is little awareness about the syndrome, even amongst health professionals, which often sends parents on a diagnostic odyssey trying to find out why their boys are different and the underlying cause remains unidentified.

Another difficulty with it is that the symptoms are highly variable, sometimes subtle and sometimes not present at all. Only about 35% present with stereotypical symptoms but up to 80% have learning difficulties and behavioural issues. Many will have developmental delays and trouble with their muscles (low muscle tone) and motor skills. Dyslexia, reading difficulties and data retrieval problems are common. A large percentage are attention deficit (ADD)

While their intellect is not compromised, their verbal IQ is below average, they often have difficulty expressing themselves. This often leads to frustration and angry outbursts. It is unclear whether some behavioural difficulties are directly caused by the syndrome or are exacerbated by early speech and learning difficulties.

KS in very young boys is especially difficult to recognise and only at puberty do some of the more obvious signs begin to show. The physical characteristic common to all KS males is that they have very small underdeveloped testicles. It is because of this that they cannot produce enough testosterone to produce sperm. This androgen masculinising hormone is also responsible for producing secondary sexual characteristics. Other more common signs are: They are often somewhat taller than genetically expected, their limbs are slightly proportionately longer. They often have knock knees (genu valgum) and high arches (pes cavus). Taurodontism (Large molar teeth with thin enamel) is common. They will have more feminine fat distribution and rudimentary breast development, (gynecomastia) high pitched voice and sparse hair. They usually have “softer more girlish faces and are generally more sensitive than most boys, they tend to be emotional and cry easily. 33% of KS males experience psychosocial issues particularly shyness and low self-esteem.

KS men also have added complications whereby they have a predisposition to developing diabetes, osteopenia and osteoporosis

It must be noted however that Klinefelter syndrome does not affect sexual orientation. When correctly treated with supplemental testosterone KS men can have normal sexual relations in adulthood.

If you suspect your son may be KS you will need to consult a geneticist. They will probably request blood tests which will include a karyotype. An endochrinologist will monitor further treatment.

Klinefelters syndrome symptoms can be mitigated by correct intervention. Testosterone supplementation must continue throughout life. This can be administered by injection or with creams or a patch. If KS is identified early there is a better chance of normal development.

A thorough psychoeducational examination will need to be conducted to determine what educational interventions are required as KS boys need assistance with the learning issues most of them have. A speech therapist can assist them and occupational therapy is recommended. Many KS boys also have motor dyspraxia.

While this all may seem alarming particularly that the syndrome is so relatively common yet somewhat unrecognised there are some positives. These are wonderful sensitive human beings. They can be encouraged that they are usually taller than average, tend to keep their youthful looks, have better intuition and emotional skills than most men and usually get on really well with women.

Further reading : www.xxy47.co.za

www.andrologyaustralia.org

www.checkyourballs.com.au

www.csvxy.org

Plus information taken from: Lawley pharmaceuticals KS info brochure 2016

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