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ABOUT US

Chromosome Disorder Outreach provides support and information to anyone diagnosed with a rare chromosome change, rearrangement or disorder. CDO actively promotes research and a positive community understanding of all chromosome disorders.

CDO is a 501c3 organization founded in 1992.

KLINEFELTER SYNDROME

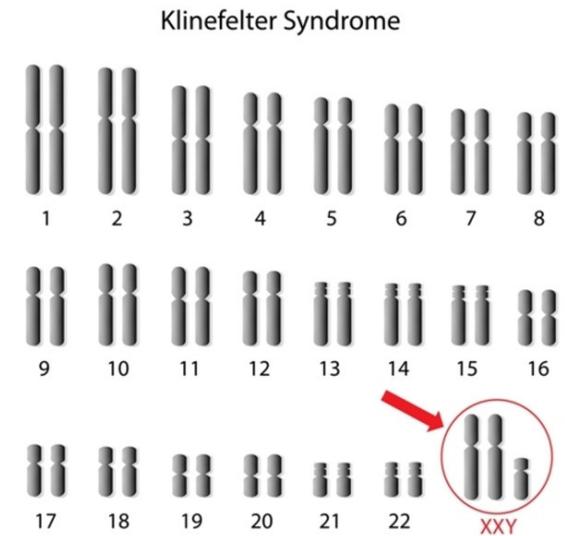
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KLINFELTER SYNDROME

Klinefelter syndrome (KS) is one of the first human disorders attributed to chromosomal abnormalities. An additional X chromosome in a man with this condition was reported in 1959. Clinically this syndrome has been known since 1942 when Dr. Harry Klinefelter and his colleagues reported a complex of hypoplastic testicles, aspermia (or low sperm count), sterility and gynecomastia (1,2).

Patients with KS usually have a 47,XXY karyotype. An additional X chromosome may be maternal or paternal; the origin of this chromosome does not change the clinical picture. KS is a common disorder; it may be found in one of 500-1000 newborn boys (1,2). In some cases 47,XXY cells may co-exist with 46,XY cells in one person, but such mosaicism is uncommon.

Sometimes patients with 3 or 4 X-chromosomes (48,XXX or 49,XXXX) are reported as variants of KS, but additional X chromosomes severely change the basic picture, and the term KS should not be used for such patients.



Newborn boys do not have specific symptoms allowing clinical diagnosis in early childhood. Most patients are diagnosed in the pubertal period or even as adults. Boys (some of them taller than their peers) have unusually small but firm testicles. They do not have usual facial hair. Bodily proportions are eunuchoid. Some patients develop gynecomastia – enlargement of breast tissue - caused by low production of testosterone. Penile size is usually normal, penile hypoplasia is mentioned only in a small proportion of affected persons.

The most common clinical symptom is sterility. Up to 3% of men examined for sterility have a 47,XXY karyotype. They produce only small amount of sperm, and in most cases there are no sperm cells in the ejaculate. However, a significant number of patients may have a small amount of sperm cells in testicular tissue, and a special procedure – testicular sperm extraction – may be used to obtain sperm cells for further in vitro fertilization procedures (3). These techniques allow a small subset of KS patients to have their own biological children. Direct transmission of KS is unknown.

KLINFELTER SYNDROME ...

continued

Another problem which may require a physician's attention is gynecomastia. Some patients need surgery to remove excessive breast tissue.

Generally patients with KS reveal some degree of developmental delay. This may be evident upon a comparison of IQ in 100 KS patients and 100 boys with a normal karyotype (4). In most individual cases however IQ is high enough to consider a person as intellectually disabled. For the majority of patients intellectual deficits are not a problem.

Because most symptoms of KS are caused by a low production of testosterone, treatment by testosterone can improve muscle mass, stimulate growth of facial and body hair and produce a more masculine appearance. If this treatment starts early the results are usually better. However, such treatment usually cannot restore normal spermatogenesis. Of course, testosterone treatment should be provided by an endocrinologist and only for those KS patients who show low testosterone level.

Children with KS have an increased risk for autism spectrum disorder and attention deficit hyperactivity disorder. In KS adults, the risk for schizophrenia and bipolar disorder is also significantly higher than for their peers with a normal 46,XY karyotype.

Patients with KS usually do not have structural abnormalities of the heart, lungs, kidneys or gastro-intestinal tract. However, it was shown that they have higher risk for cerebro-vascular diseases (5). As a result the mean life expectancy for KS patients is about 5 years less than the life expectancy for 46,XY persons.