



**ÖSTERREICHISCHE KLINEFELTER  
SYNDROM GRUPPE**  
SHG SALZBURG



Englisch

**SHG KLINEFELTER-SYNDROM**

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The syndrome of Klinefelter

The syndrome of Klinefelter is a genetic anomaly characterized by the presence of an additional X chromosome at individuals of male sex. The chromosomal formula of the subject is thus 47XXY instead of 46XY. All the cells are not always affected (mosaic form).

The syndrome is named according to the American doctor Dr. Harry F. Klinefelter, who, in 1942, described for the first time the characteristics. In 1959 the bond between the presence of a supernumerary chromosome and the symptoms was discovered.

The anomaly is due to a nondisjunction of the chromosomes during the formation of the ovules or the spermatozoa. It is about a spontaneous phenomenon which occurs without apparent reason and of which, in theory, each one can be concerned.

About 10.000 boys and men reached of the syndrome live in Austria.

The various physical and mental symptoms can strongly vary in width between the individuals. Part of the symptoms can be avoided, eliminated or be attenuated by a medical treatment or psychological measurements - teaching.

Consequences of the syndrome of Klinefelter

The underdevelopment of the testicles leads to an infertility which may not be treated and nonreversible, while the sexual life can remain normal well. As the testicles are very small, the testosterone is produced only in small quantity. This causes a puberty late or even absent, a facial and body pilosity reduced, as well as the presence of developed centres. The subjects are in addition often of big size.

The boys carrying the syndrome of Klinefelter are often babies tranquil and generally more passive than their brothers and sisters. More often than among normal boys of the delays of development, disorders of training and communication (especially on the level of the language) are observed.

Instructions qualified for the parents and the teachers help to avoid certain mental and school problems.

The treatment consists of a hormonothérapie containing testosterone as of puberty. In substituent the testosterone, considerable symptoms can be reduced and the quality of life is appreciably improved. The appearance of osteoporosis, caused by the hormonal lack, is thus avoided. Various preparations containing testosterone are available for the hormonothérapie.

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