

47, XXY (KS) is not inherited. Males with 47, XXY (KS) have one extra X chromosome because of a nondisjunction error that randomly occurs during the division of the sex chromosomes in the egg or sperm. Some males with 47, XXY (KS) are mosaic, meaning that some cells have an extra X chromosome and other cells do not. Mosaic 47, XXY (KS) occurs because of an error in the division of the sex chromosomes in the zygote after fertilization. The extra X chromosome typically results in primary testicular failure leading to androgen deficiency. 47, XXY (KS) is the most common human sex chromosome disorder and occurs in approximately 1 in 500-1,000 males. It is estimated that 3,000 affected boys are born each year in the United States. Kallmann syndrome is a rare inherited disorder that mostly, but not exclusively, affects men. The major characteristics of Kallmann syndrome, in both men and women, are the failure to experience puberty and the complete or partial loss of the sense of smell. Failure to go through puberty reflects a hormonal imbalance that is caused by a failure of a part of the brain known as the hypothalamus. Patients with Kallmann syndrome show evidence of small genitalia, sterile gonads that cannot produce the sex cells (hypogonadism), and a loss of the sense of smell (anosmia). The impaired production of hormones as well as sperm and egg cells causes delayed puberty, growth and infertility. (For more information on this disorder, choose "Kallmann syndrome" as your search term in the Rare Disease Database.) Males with 47, XXY (KS) are most commonly identified before birth (e.g. through prenatal screenings for chromosomal disorders), at puberty, or later in life because of infertility. 47, XXY (KS) is diagnosed by a chromosome karyotype analysis on a blood sample or by a chromosomal microarray (CMA) test. CMA consists of an oral cheek (buccal) swab and is an easy and painless way to detect abnormalities of chromosome numbers and provide a definitive diagnosis. 47, XXY (KS) can also be diagnosed prenatally on chorionic villous or amniotic fluid cells.