



## Sex chromosome aneuploidies

Very variable presentations, usually under-diagnosed, clinical interpretation of prenatal diagnosis is limited

The detection rate of sex chromosome aneuploidies is 99.6%

Syndromes	Definition	Prevalence	Description	Management	Positive predictive value of a positive screening test result
<b>Turner syndrome (45,X)</b>	<p><b><u>Affected : female individuals</u></b></p> <p>- Complete or partial absence of the X chromosome, frequent mosaic forms (45, X/46,XX)</p>	About 1 in 1800 girls	<p><b>Great variability in expression and severity</b> (some affected pregnancies will not reach term, while some women may be under-diagnosed because they are virtually asymptomatic).</p> <p><b><u>The 3 main characteristics</u></b></p> <p>-Smaller stature -Impaired ovarian function -Infertility (early ovarian failure; if reserve too low, possibility of egg donation)</p> <p><b><u>Very frequent characteristics (&gt;50%)</u></b></p> <p>Lymphatic edema (may manifest as nuchal hyperclarity or cystic hygroma on first trimester ultrasound), low capillary implantation, webbed neck, tendency to obesity</p> <p><b><u>Frequent characteristics (5-50%)</u></b></p> <p>Learning disability, heart and/or kidney defects, hearing impairment, hypertension, hypothyroidism, glucose intolerance or diabetes</p>	<p>- <u>If fetal diagnosis</u> : recommendation for detailed ultrasound and echocardiogram</p> <p>- <u>For an affected individual</u> : Growth hormone, hormone replacement therapy, cardiology follow-up (particularly if pregnancy is planned)</p>	30% ; low due to higher number of placental mosaicism

<p><b>Klinefelter syndrome (47,XXY)</b></p>	<p><b><u>Affected: male individuals</u></b> - The presence of an additional X chromosome</p>	<p>About 1 in 600 boys</p>	<p>- <b>A large number of affected individuals are under diagnosed as almost asymptomatic.</b> - Increased risk of learning disabilities, delayed speech, social, emotional and behavioral difficulties - Can be taller than average - Mammary gland may be prominent (gynecomastia), often testicles are small; produce less testosterone - Infertility (non-obstructive azoospermia, sometimes possibility of IVF with testicular sperm extraction versus the use of sperm donation)</p>	<p>- <u>If fetal diagnosis:</u> standard follow-up - <u>For an affected individual :</u> - Testosterone - Developmental intervention if symptomatic (e.g. speech therapy) - In adulthood, fertility treatment (assisted reproduction)</p>	<p>74%</p>
<p><b>Triple X syndrome (47,XXX)</b></p>	<p><b><u>Affected : female individuals</u></b> - The presence of a supernumerary X chromosome</p>	<p>About 1 in 1000 girls</p>	<p>- <b>Most people are not diagnosed as asymptomatic</b> - Increased risk of learning disabilities, delayed speech and language development, behavioral difficulties, emotional difficulties - Can be taller than average</p>	<p>- <u>If fetal diagnosis:</u> standard follow-up - <u>For an affected individual :</u> Developmental intervention if symptomatic (e.g. speech therapy)</p>	<p>54%</p>
<p><b>Jacobs syndrome (47,XYY)</b></p>	<p><b><u>Affected : male individuals</u></b> - The presence of an extra Y chromosome</p>	<p>About 1 in 1000 boys</p>	<p>- <b>Most people are not diagnosed as asymptomatic</b> - Increased risk of learning disabilities, delayed speech and language development, behavioral difficulties, emotional difficulties - Can be taller than average</p>	<p>- <u>If fetal diagnosis:</u> standard follow-up - <u>For an affected individual :</u> Developmental intervention if symptomatic (e.g. speech therapy)</p>	<p>75%</p>