Chromosomal Abnormalities Pamphlet

Autosomal Abnormalities (chromosomes #1 - 22)

Nine percent of spontaneous abortions (miscarriages) are caused by trisomy 13, 18, or 21; but 0.1% of newborns have these trisomies.

Down Syndrome

Down syndrome is trisomy 21 (3 copies of chromosome 21 instead of 2 copies → this means the person has 47 chromosomes instead of 46 chromosomes). It is characterized by mental retardation, an abnormal pattern of palm creases, a flat face, sparse, straight hair, and short stature. People with Down syndrome have a high risk of having cardiac anomalies, leukemia, cataracts, and digestive blockages.

Life expectancy of Down syndrome individuals is in the middle teens but some live much longer.

The gene responsible for Alzheimer’s is on chromosome 21. Down’s are at increased risk for developing Alzheimer’s.

Down Syndrome is associated with maternal age. Older women, particularly those older than 40, are more likely to have a Down Syndrome child.

Translocation Down Syndrome

A translocation is the movement of a chromosomal segment from one chromosome to another nonhomologous chromosome.

Five percent of Down Syndrome cases involve a translocation.

The translocation often involves chromosome 14.

In the translocation diagrammed below, chromosome #21 has become fused with chromosome #14.

During meiosis, the two chromosomes might align so that each daughter cell receives one chromosome 21 as shown below. This will produce a normal egg.
If the chromosomes align as illustrated below, one daughter cell will receive two chromosome 21s and the other will not receive any. When a gamete with two 21s fuses with a normal gamete, the result is a zygote with three chromosome 21s.

This form runs in families and is not age-related.

**Trisomy 18 (Edward Syndrome)**

Trisomy 18 is associated with mental and physical retardation, skull and facial abnormalities, defects in all organ systems, and poor muscle tone.

Mean survival is 2 to 4 months.

**Deletions**

Deletions are fragments of chromosomes that are missing. They are usually lethal when homozygous and cause abnormalities when heterozygous.

Radiation, viruses, chemicals, and unequal crossing-over may cause them.

**Cri du Chat Syndrome**

Cri du chat syndrome is due to a deletion of a portion of chromosome 5. Cri du chat individuals are mentally retarded. "Cri du chat" is French for "cry of the cat". The infants cry sounds like a cat. Death usually occurs in infancy or early childhood.

**Abnormalities of the Sex Chromosomes (chromosome “pair” 23)**

**Turner Syndrome - XO**

Females with Turner syndrome have monosomy for their sex chromosomes. They lack a 2nd sex chromosome – the missing chromosome is designated by a 0 (zero). As a result they have 45 chromosomes instead of 46 chromosomes.

Characteristics of Turner syndrome include the following:

Sexually underdeveloped
Short stature
Folds of skin on the back of the neck
Wide-spaced nipples
Narrow aorta
Pigmented moles
97% die before birth
Malformed elbows
Infertile
Normal Intelligence

The incidence of Turner syndrome is 1 in 2000 female births.

Turner syndrome individuals that are treated with hormones lead fairly normal lives.

**XXX - Triple-X Syndrome (also XXXY and XXXXX)**

Triple-X individuals are tall and thin and have menstrual irregularities. Their IQ is in the normal range but it is slightly reduced.

The incidence of Triple-X Syndrome is 1 in 1,500 female births.

Additional X chromosomes are associated with an increased mental handicap.

**XXY - Klinefelter Syndrome (also XXXY)**

Males with two or more X chromosomes have Klinefelter Syndrome.

The incidence of Klinefelter Syndrome is 1 in 1000 male births.

Symptoms include reduced sexual maturity and secondary sexual characteristics, breast swelling, and no sperm. Klinefelter males are slow to learn and individuals with additional X’s (XXXXX) may be mentally retarded.

**XYY - Jacob Syndrome**

XYY males are tall, have acne, speech, and reading problems.

Although there are a disproportionate number in penal institutions (prison), 96% of Jacob’s Syndrome men are normal.