**EXPLANATION OF CHROMOSOME DISORDERS**

**Cri-du-chat**

Babies with the "cry of the cat" syndrome have a cry which sounds like that of a cat in distress because the infant's larynx is improperly developed. The cause of this condition is a deletion of about half of the short arm of chromosome number five. Cri-du-chat babies are severely mentally retarded, have a small cranium, a small jaw and a moon-shaped face. The incidence of this syndrome is 1/100,000 live births.

Karyotype: 46XX or 46XY with one chromosome #5 upper arm deletion

**Down Syndrome**

Trisomy 21, one of the most common causes of mental retardation is due to an extra chromosome 21. This results in a number of characteristic features, such as short stature, broad hands, stubby fingers and toes, a wide rounded face, a large protruding tongue that makes speech difficult and mental retardation. Individuals with this syndrome have a high incidence of respiratory infections, heart defects and leukemia. The average risk of having a child with trisomy 21 is 1/750 live births. Mothers in their early twenties have a risk of 1/1,500 and women over 35 have a risk factor of 1/70, which jumps to 1/25 for women 45 or older.

Karyotype: 47XX or 47XY with 3 of the chromosome #21

**Edward's Syndrome**

This syndrome, trisomy 18 (an extra chromosome 18), produces severe mental retardation and a highly characteristic pattern of malformations such as elongated skull, a very narrow pelvis, rocker bottom feet, malformed heart and a grasping of the two central fingers by the thumb and little finger. In addition, the ears are often low set and the mouth and teeth are small. Nearly all babies born with this condition die in early infancy. The frequency of this syndrome is 1/5,000 live births.

Karyotype: 47XX or 47XY with 3 of the chromosome #18

**Jacobs**

A chromosome abnormality which is caused by nondisjunction (non-separation) of the Y chromosome during the second phase of meiosis. This condition only occurs in men and happens when they have an extra Y chromosome. Men with this abnormality are tall, develop heavy cases of acne and have low mental ability. Occurrence is 1/1,000 live male births. Karyotype: 47XYY (extra sex chromosome - extra Y)

**Klinefelter Syndrome**

A condition occurring in 1/1,000 male live births. Characteristics associated with this condition are tall stature, small testicles, developed breasts, sterility and mental deficiency. Most men with this syndrome appear normal in other ways. This syndrome only occurs in men and affects the sex chromosomes.

Karyotype: 47XXY or 47XXXY (extra sex chromosomes)

**Patau Syndrome**

This syndrome (trisomy 13 - extra chromosome 13) causes severely abnormal cerebral functions and virtually leads to death in early infancy. The baby has very pronounced clefts of the lip and palate, broad nose, polydactyly (extra fingers and toes), small cranium and nonfunctional eyes. Heart defects and severe mental retardation are also part of the clinical picture. The frequency is 1/15,000 live births.

Karyotype: 47XX or 47XY with 3 of the chromosome 13

**Triple X**

Superfemale (XXX). Occurs at a frequency of 1/1,000 female live births. No specific abnormalities are associated with this condition. The vast majority of women who have this condition are normal mentally, but may have underdeveloped genitalia and limited fertility. They also may have neuromotor delays. This condition only occurs in females and affects the sex chromosomes.

Karyotype: 47XXX (affects sex chromosomes - extra X chromosome)

**Turner Syndrome**

This condition happens when an X-carrying sperm fertilizes an ovum that lacks an X, or when a sperm lacking an X or Y chromosome fertilizes an X-bearing egg. This syndrome only affects girls and causes them to be missing an X chromosome (XC). These girls appear to be normal before puberty, although they are shorter and have a chunky build. At birth, the distinguishable characteristics include a webbed neck. At sexual maturity, the secondary sex characteristics are not developed. There also is no menstruation or breast development and they are usually sterile. The frequency is 1/2,500 live female births. -- Karyotype: 45X or 45XO (affects sex chromosomes - missing an X)