**Genetic Disorder Round-Up**

**Down Syndrome:**

Down syndrome is a genetic disorder caused by extra genetic material. It affects over 350,000 people in the United States alone and is the most common (1 in 800 live births) imbalance in the number of autosomes in people. The effects of Down syndrome vary greatly from person to person but can include mental impairments, eyes that slant upward, and heart defects.

People with Down syndrome have thee copies of chromosome 21. For this reason, Down syndrome is also called trisomy 21. Where does the extra chromosome come from? In 90% of the cases, the additional chromosome comes from thee mother’s egg. This increases when the mother is over 35 years of age.

**Turner Syndrome:**

Turner syndrome affects 60,000 girls and woman in the United States. This disorder occurs in 1 in 2000 to 1 in 2500 live births, with about 800 new cases diagnosed each year. Symptoms include short stature, lack of ovarian development, webbed neck arms that turn out slightly, and a low hairline in the back of the head. Heart defects can also occur. Most are sterile.

Woman with Turner syndrome have only 1 copy of the X chromosome. This is an example of monosomy. Where does the single X come from? In 75 to 80% of the cases, the single X comes from the mother’s egg because the father’s sperm cell that fertilizes the egg is missing a sex chromosome.

**Klinefelter Syndrome:**

Klinefelter syndrome occurs in 1 in 500 to 1 in 1000 live births. People with this disorder develop as males with subtle characteristics that become apparent during puberty. Males are often tall and usually do not develop secondary sex characteristics such as facial hair, underarm hair, or pubic hair. Most males are sterile and have smaller than normal testicles. These men commonly develop slightly enlarged breasts. Some males develop an increased risk of breast cancer.

Men and boys with Klinefelter syndrome have a Y chromosome and 2 X chromosomes. This is an example of trisomy. Where does the extra chromosome come from? In about half of the cases, the extra X chromosome comes from the mother’s egg, while the other half of the cases, the extra chromosome comes from the father’s sperm.

**Patau Syndrome:**

Patau syndrome is also called trisomy 13. Characteristics of this condition are severe mental impairment, small eyes that may exhibit a split in the iris, a cleft lip and/or palate, weak muscle tone, an increased risk of heart defects, skeletal abnormalities, facial malformation, and other medical problems. Affected individuals rarely live past infancy because of the life threatening medical problems associated with this condition. Trisomy 13 affects about 1 in 10,000 live births. The risks of having a child with trisomy 13 increase as a woman gets older.

People with trisomy 13 have additional DNA from chromosome 13 in their cells. The extra material disrupts the normal course of development. Most cases of trisomy 13 result when each cell in the body has 3 copies of chromosome 13 instead of the normal 2 copies. In other case, only some of the cells have the extra copy of chromosome 13. In other cases, part of chromosome 13 becomes attached to another chromosome (translocated) before or at conception. With a translocation, the person has a partial trisomy for chromosome 13 and often the physical signs of the syndrome differ from those typically seen in trisomy 13.

**Edwards Syndrome:**

This is a genetic disorder that an extra copy of the chromosome is present in the 18th chromosomal pairing. This is the second most common form of genetic disorders in humans. Edwards syndrome occurs in 1 in 5000 live births.

Mental impairment is an inevitable result of this condition. Many children die within a very short time after birth. There is also an increased risk of death occurring in utero. Very few trisomy 18 children live beyond one year of age. Characteristics of this syndrome are low set ears, deformed fingers, narrow nose, and a receding jaw as well as congenital heart disease.

**XYY Syndrome:**

XYY syndrome can also be called YY syndrome or Jacob’s syndrome. Affected males have an extra Y chromosome. Affected individuals are usually very tall. Many experience severe acne during adolescence. Additional symptoms may include learning disabilities and behavioral problems such as impulsivity. Intelligence is usually in the normal range, although IQ is on average 10-15 points lower than siblings. Many have delayed motor skills, such as crawling or walking, low muscle tone, speech impairments, low testosterone levels, and low sperm counts that can lead to infertility. Men with this syndrome appear normal physically.

**Triple X Syndrome:**

Triple X syndrome, also called trisomy X, is characterized by the presence of an additional X chromosome in each of a female's cells. Although females with this condition may be taller than average, this chromosomal change typically causes no unusual physical features. Most females with triple X syndrome have normal sexual development and are able to conceive children.

Triple X syndrome is associated with an increased risk of learning disabilities and delayed development of speech and language skills. Delayed development of motor skills (such as sitting and walking), weak muscle tone, and behavioral and emotional difficulties are also possible, but these characteristics vary widely among affected girls and women. Seizures or kidney abnormalities occur in about 10 percent of affected females.

This condition occurs in about 1 in 1,000 newborn girls. Five to 10 girls with triple X syndrome are born in the United States each day.