FACT SHEET

Down Syndrome

What is Down Syndrome?

Down Syndrome is a genetic condition that causes delays in physical and intellectual development. It occurs in approximately one in every 800 live births. Individuals with Down Syndrome have 47 chromosomes instead of the usual 46.

Three genetic variations can cause Down Syndrome. Approximately 92% of the time, Down Syndrome is caused by the presence of an extra **Chromosome 21** in all cells of the individual. In such cases, the extra chromosome originates in the development of either the egg or the sperm. Consequently, when the egg and sperm unite to form the fertilized egg, three-rather than two-Chromosomes 21's are present. As the embryo develops, the extra chromosome is repeated in every cell. This condition, in which three copies of Chromosome 21 are present in all cells of the individual, is called **Trisomy 21**.

In approximately 2-4% of cases, Down Syndrome is due to **Mosaic Trisomy 21**. This situation is similar to simple Trisomy 21, but, in this instance, the extra Chromosome 21 is present in some, but not all, cells of the individual. For example, the fertilized egg may have the right number of chromosomes, but, due to an error in chromosome division early in embryonic development, some cells acquire an extra Chromosome 21. Thus, an individual with Down Syndrome due to Mosaic Trisomy 21 will typically have 46 chromosomes in some cells, but will have 47 chromosomes (including an extra Chromosome 21) in others. In this situation, the range of the physical problems may vary, depending on the proportion of cells that carry the additional Chromosome 21.

How is it manifested?

- muscle hypotonia, low muscle tone
- flat facial profile, a somewhat depressed nasal bridge and a small nose
- oblique palpebral fissures, an upward slant to the eyes
- dysplastic ear, an abnormal shape of the ear
- a single deep crease across the center of the palm
- hyperflexibility, an excessive ability to extend the joints
- epicanthal folds, small skin folds on the inner corner of the eyes
- excessive space between large and second toe
- enlargement of tongue in relationship to size of mouth

Who is affected?

It is estimated that 1 in every 800-1,000 children born in Canada are diagnosed with Down Syndrome. It is the most frequently occurring chromosomal abnormality.

How is it diagnosed or detected?

Down Syndrome is usually identified at birth or shortly thereafter. Initially, the diagnosis is based on physical characteristics that are commonly seen in babies with Down Syndrome. These include low muscle tone, a single crease across the palm of the hand, a slightly flattened facial profile, and an upward slant to the eyes. The diagnosis must be confirmed by a chromosome study (karyotype). A karyotype provides a visual display of the chromosomes grouped by their size, number, and shape. Chromosomes may be studied by examining blood or tissue cells.

Additional Resources:

DOWN SYNDROME ASSOCIATION OF TORONTO – www.dsat.ca
Down Syndrome Association of Toronto is a non-profit organization providing support and information to parents of children with Down Syndrome, students and teachers.

CANADIAN DOWN SYNDROME SOCIETY – www.cdss.ca
The Canadian Down Syndrome Society (CDSS) is a resource linking parents and professionals through advocacy, education and providing information.