



Down Syndrome Association of Toronto
40 Wynford Drive Suite 303
Toronto, ON M3C 1J5
416-966-0990
www.dsat.ca

Down syndrome - What is It?

Down syndrome is a chromosomal disorder that occurs in approximately 1 in 700 live births in Canada. Although it is not known for sure, it is possible that an error in cell division causes this extra chromosome. This extra chromosome material was present at conception, either from the father's sperm or the mother's egg, each of which contributes 23 chromosomes. A person with Down syndrome will have 47 chromosomes in each cell instead of 46. It is most important to know that nothing the mother did during pregnancy could have caused this to happen.

How did our doctor suspect that our baby has Down syndrome?

Trisomy for the segment of chromosome 21q22 produces the mental and physical characteristics of Down syndrome. The most common characteristics used for diagnosis of Down syndrome in newborns are:

1. Low muscle tone
2. Low nasal bridge, small nose and ears
3. Eye openings slant upwards
4. A ring of white dots in the iris of the eye
5. A single crease across the centre of the palm.
6. Excessive ability for extending extremities
7. Only one flexion furrow on fifth finger instead of two or, creases close together

Factors affecting the development of a child with Down syndrome

We recognize that there is a range of development in children with Down syndrome from severe delay to near normal. Factors influencing this range, in addition to early intervention programs, include the other genes the child inherited from his family, effects of fetal distress from a difficult birth, and health problems resulting in low oxygen or prolonged hospitalization for health reasons. Some children with Down syndrome are described as mosaic. This means they carry some normal cells and some Down syndrome cells in their body. Depending upon the distribution of normal cells, the fact that there are some Down syndrome cells may go undetected by routine chromosome studies. Children who are "mosaic" often achieve more normal levels of development.

Who has the highest risk of having a child with Down syndrome?

For women over 35, the risk of having a child with Down syndrome is significantly increased (age 35: 1 in 350). At age 40 the risk increases to 1 in 100 births of a child having Down syndrome. At age 45 the risk

becomes approximately 1 in 25. Since many couples are postponing parenting until the 4th decade of life, the incidence of Down syndrome is expected to increase.

Therefore, genetic counselling for parents is becoming increasingly important. Yet many physicians are still not fully informed about how to advise patients about the risk of Down syndrome, advances in diagnosis, and the progress in care and treatment of those born with Down syndrome.