What is Down syndrome?

Down syndrome is a genetic chromosomal disorder that occurs in approximately one of 830 live births.

What are the chromosome basics of Down syndrome?

Genes on an extra copy of chromosome 21 are responsible for all characteristics associated with Down syndrome. Normally, each human cell contains 23 pairs of different chromosomes. Each chromosome carries genes, which are needed for proper development and maintenance of our bodies. At conception, an individual inherits 23 chromosomes from the mother (through the egg cell) and 23 chromosomes from the father (through the sperm cell).

However, sometimes a person inherits an extra chromosome from one of the parents. In Down syndrome, an individual most often inherits two copies of chromosome 21 from the mother and one chromosome 21 from the father for a total of three chromosomes 21. Because Down syndrome is caused by the inheritance of three chromosomes 21, the disorder is also called trisomy 21. About 95 percent of individuals with Down syndrome inherit an entire extra chromosome 21.

What are the risk factors for conceiving a child with Down syndrome?

The only well known risk factor for conceiving a child with Down syndrome is advanced maternal age. The older the woman is at conception, the greater the risk of having a child with Down syndrome.

Parents who have conceived a child with Down syndrome have a 1 percent increased risk of conceiving another child with Down syndrome. If a parent is a carrier of a chromosome 21 translocation, the risk increases. Women with Down syndrome have a 50 percent risk of conceiving a child with Down syndrome. If the father has Down syndrome, the risk of conceiving a child with Down syndrome is also increased.

What are the characteristic features and symptoms of Down syndrome?

Although the severity of Down syndrome ranges from mild to severe, most individuals with Down syndrome have widely recognizable physical characteristics. These include:

- flattened face and nose, a short neck, a small mouth sometimes with a large, protruding tongue, small ears, upward slanting eyes that may have small skin folds at the inner corner (epicanthal fold);
- white spots (also known as Brushfield spots) present on the colored part of the eye (iris);
- hands that are short and broad with short fingers and with a single crease in the palm, first and second toes widely separated, increased skin creases;
- poor muscle tone and loose ligaments; and
• delayed development and growth, lesser than average height, failure to read, developmental milestones are not reached.

**What about cognitive impairment in Down syndrome?**

Down syndrome is the leading cause of cognitive impairment (problems with thinking and learning). Cognitive development is often delayed, and all individuals with Down syndrome have mild to severe learning difficulties that last throughout their lives. How the extra chromosome 21 leads to cognitive impairment is not entirely clear. The average brain size of a person with Down syndrome is small, and scientists have found alterations in the structure and function of certain brain areas such as the hippocampus and cerebellum. Particularly affected is the hippocampus, which is responsible for learning and memory. Scientists are using human studies and animal models of Down syndrome to find out which specific genes on the extra chromosome 21 lead to different aspects of cognitive impairment.

**What other conditions are associated with Down syndrome?**

Heart conditions: Apart from cognitive impairment, the most common medical conditions associated with Down syndrome are congenital heart defects. About half of all people with Down syndrome are born with a heart defect. Some babies will require surgery shortly after birth to correct these heart defects.

Gastrointestinal conditions: Gastrointestinal conditions are also commonly associated with Down syndrome, especially esophageal atresia, tracheoesophageal fistula, duodenal atresia or stenosis, Hirschsprung disease, and imperforate anus. Individuals with Down syndrome are at a higher risk for developing celiac disease. Corrective surgery is sometimes necessary for gastrointestinal problems.

Cancer: Certain types of cancer are more frequently found in Down syndrome, such as acute lymphoblastic leukemia (a type of blood cancer), myeloid leukemia and testicular cancer. Solid tumors, on the other hand, rarely occur in this population.

Other medical conditions include: hearing loss, frequent ear infections (otitis media), underactive thyroid (hypothyroidism), cervical spine instability, visual impairment, sleep apnea, obesity, constipation, infantile spasms, seizures, dementia and early-onset Alzheimer's disease.

About half of individuals with Down syndrome have coexisting psychiatric or behavior conditions, such as: autism spectrum disorders, attention deficit hyperactivity disorder (ADHD), depression, stereotypical movement disorders and obsessive compulsive disorder.

**How is Down syndrome managed?**

Although the genetic cause of Down syndrome is known, there is currently no cure. Due to advances in technology, scientists are slowly beginning to understand which genes present in three copies are responsible for particular Down syndrome characteristics, but it will take many years to fully grasp the complex interplay between the different genes. Much research to date is focused on understanding the causes of impaired cognition in Down syndrome and on finding potential therapies that might improve learning.

Corrective surgery for heart defects, gastrointestinal irregularities and other health issues is necessary for some individuals. Regular health checkups should be scheduled to screen for other conditions, such as visual impairments, ear infections, hearing loss, hypothyroidism, obesity and other medical conditions.

**What about early intervention and education for Down syndrome?**

As with all babies, it is very important to stimulate, encourage and educate children with Down syndrome from infancy. Programs for young children with special needs are offered in many communities. Early intervention programs, including physical therapy, occupational therapy and speech therapy, can be very helpful.
What are the needs of infants and preschool children with Down syndrome?

Like all children, children with Down syndrome greatly benefit from being able to learn and explore in a safe and supportive environment. Being included in family, community and preschool life will help a child with Down syndrome develop to his or her full potential.

While social development and social learning are often quite good, development in other areas such as motor skills, speech and language are usually delayed. Many children with Down syndrome eventually reach most developmental milestones, but mild to severe learning difficulties will persist throughout life.

In general, children with Down syndrome are more prone to illnesses and vision and hearing impairments, which can contribute to the child's learning difficulties. Regular health checkups are very important. Some children may have more severe developmental delays. This could be due to coexisting medical or psychiatric conditions, such as seizures, autism or ADHD.

RESOURCES:

6. PA Special Kids Network - [http://www.health.state.pa.us/skn](http://www.health.state.pa.us/skn)
8. PA Department of Health - [http://www.helpinpa.state.pa.us/HelpInPA/agencysearch.aspx](http://www.helpinpa.state.pa.us/HelpInPA/agencysearch.aspx)