

What is Down syndrome?

Down syndrome is a genetic chromosomal condition that occurs in approximately one of 700 live births.

What are the chromosome basics of Down syndrome?

Typically, each human cell contains 23 pairs of different chromosomes for a total of 46 chromosomes. Each chromosome carries genes, which are needed for proper development, function, and maintenance of our bodies. At conception, an individual usually 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 95% of people with Down syndrome, the individual inherits two copies of chromosome 21 from one parent and one copy of chromosome 21 from the other parent, for a total of three chromosomes 21. Because Down syndrome is caused by the inheritance of three chromosomes 21, the condition can also be called Trisomy 21. 3-4% of people with Down syndrome inherit an extra chromosome 21 through a process called translocation. This is important to know because the translocation type of Down syndrome can potentially have an increased chance of happening again in a future pregnancy. The remaining 1-2% of people with Down syndrome inherit an extra chromosome 21 in only a portion of their cells through a process called mosaicism.

What are the chances of conceiving a child with Down syndrome?

The only known factor that increases the chances of conceiving a child with Down syndrome is advanced maternal age. The older the woman is at conception, the greater the chance of having a child with Down syndrome.

Parents who have conceived a previous child with Down syndrome are felt to have a 1 percent chance of conceiving another child with Down syndrome. As mentioned earlier, if the previous child with Down syndrome has the translocation type, the chances can be much higher in future pregnancies since one of the parents may carry a chromosome 21 translocation themselves.

What are the characteristic features and symptoms of Down syndrome?

Individuals with Down syndrome typically can have certain 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 on the feet;
- low muscle tone and loose ligaments; and
- delayed development and growth, smaller than typical height.

What about cognitive impairment in Down syndrome?

Cognitive development is varied, but most individuals with Down syndrome have mild to moderate learning difficulties that can last throughout their lives. How the extra chromosome 21 leads to cognitive difficulties is not entirely clear. Scientists are using human studies and animal models of Down syndrome to find out which specific genes on the extra chromosome 21 may lead to different aspects of cognitive development.

What other conditions are associated with Down syndrome?

Heart conditions: A common medical condition associated with Down syndrome is a congenital heart defect. 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. Depending on the specific heart defect, they may also resolve on their own.

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. Corrective surgery is sometimes necessary for gastrointestinal problems.

Cancer: Certain types of cancer are found more often in people with Down syndrome, such as leukemia. Solid tumors, on the other hand, rarely occur in this population.

Other possible medical conditions include hearing loss, frequent ear infections (otitis media), underactive thyroid (hypothyroidism), visual impairment, constipation, or a specific type of seizure called infantile spasms..

Some individuals with Down syndrome can develop psychiatric or behavior conditions, such as autism spectrum disorders, attention deficit hyperactivity disorder (ADHD), depression, stereotypical movement disorders or obsessive compulsive disorder. These can be managed similarly as they are in other children.

How is Down syndrome managed?

In many ways, babies with Down syndrome are just like other babies. All babies need to be fed, have their diapers changed, and have playtime, but, most of all, they need to be loved. This is the same for a baby with Down syndrome.

Infants with Down syndrome need to be followed by a primary care provider just like other children for immunizations as well monitoring for growth and development. There are healthcare guidelines from the American Academy of Pediatrics specifically pertaining to the needs of children with Down syndrome (see resources section). Children with Down syndrome will need to have certain medical follow-ups (Cardiology, Ophthalmology, Audiology, etc) and lab testing and particular intervals. Depending on their individual health needs, infants with Down syndrome may sometimes require corrective surgery for heart defects or gastrointestinal issues early in life.

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 can be delayed. Many children with Down syndrome eventually reach their developmental milestones, but mild to moderate learning difficulties may persist.

In general, most children with Down syndrome do well with proper medical care and developmental support services. Regular health checkups are very important. Medical providers should pay particular attention to following the AAP Healthcare Guidelines. Whenever possible, a regional Down Syndrome Center should be involved to provide expert medical guidance and advocate for the children to reach their full potential.

RESOURCES:

1. Down Syndrome Center of Western Pennsylvania - <http://www.chp.edu/CHP/downsyndrome>
2. Trisomy 21 Program, Children's Hospital of Philadelphia - <http://www.chop.edu/centers-programs/trisomy-21-program>
3. Dear Self About Down Syndrome Parent blog – <http://dsaboutds.wordpress.com>
4. Down Syndrome Center Podcast – <http://www.chp.edu/dscpodcast>
5. National Down Syndrome Society - <http://www.ndss.org/>
6. National Down Syndrome Congress - <http://www.ndscenter.org>
7. Centers for Disease Control - <http://www.cdc.gov/ncbddd/birthdefects/downsyndrome.html>
8. American Academy of Pediatrics Healthcare Guidelines (parent version) – <https://www.healthychildren.org/English/health-issues/conditions/developmental-disabilities/Pages/Children-with-Down-Syndrome-Health-Care-Information-for-Families.aspx>
9. American Academy of Pediatrics Healthcare Guidelines (medical version) - <http://pediatrics.aappublications.org/content/128/2/393.full.pdf+html?sid=beb7145a-35b2-46b7-979f-ad860fe639d6>
10. PA Special Kids Network - <http://www.health.state.pa.us/skn>