

Down Syndrome

Also known as: Trisomy 21

Clinical Characteristics

Down syndrome is one of the more well-known genetic syndromes, and is the most frequent known cause of mental retardation. The characteristics of the syndrome are widely known, including a recognizable facial appearance, short stature, hypotonia, and some degree of mental retardation. Children with Down syndrome are at increased risk for congenital heart disease, intestinal tract obstructions, respiratory problems, skeletal problems, epilepsy, leukemia, abnormal thyroid function, and middle ear problems that may lead to hearing loss. The typical facial appearance includes upslanted eyes, a wide nasal bridge, white brushfield spots in the eyes, small ears, and a small mouth with a relatively large and protruding tongue. Down syndrome can affect nearly any of the body's systems; some characteristics are simply more common than others.

Down syndrome and Hearing Loss

Approximately 40-70% of individuals with Down syndrome have middle ear problems. Eustachian tube dysfunction seems to be the primary cause of hearing loss; fluid accumulates in the middle ear, leading to recurrent otitis media with effusion. Hearing loss may also be caused by blockages in the narrow ear canals. This and sinusitis can be problematic when attempting to effectively diagnose and treat an individual's hearing loss. A small minority of individuals have permanent sensorineural deafness.

Natural History

Down syndrome is diagnosed in all ethnicities and in both males and females with a slight male preponderance. The prevalence of Down syndrome at birth is estimated to be between 1 in 650 and 1 in 700. That number decreases to 1 in 1,000 by the age of 12 months, as some infants die during the neonatal period.

During infancy, the most serious issues for a baby with Down syndrome are heart disease and duodenal atresia. At least half of babies have a cardiac defect, the most common of which is an atrioventricular septal defect. Babies are often "floppy" due to hypotonia, but otherwise tend to develop normally during the first few years. By age 2 or 3 years, though, developmental delay becomes apparent. Eventually, all children with Down syndrome will be classified as mentally retarded. Growth is generally slow and requires the use of specific growth charts.

Hearing loss (in about 75%), otitis media (50-75%), strabismus and other eye abnormalities (60%), obstructive sleep apnea (50-75%), cardiac problems (50%) hypothyroidism (15%), digestive complications (5%), leukemia (<1%), respiratory tract infections, and atlantoaxial instability are all potential issues during childhood. Hearing loss and hypothyroidism concerns continue into adulthood. Adults with Down syndrome

may also be affected by celiac disease, obesity, immunologic concerns, seizure disorders, and hip degeneration. Many of these health issues can be successfully treated with proper interventions. In general, individuals with Down syndrome do not reproduce.

Most individuals with Down syndrome fall into the mild to moderate range of mental retardation, capable of reading, writing, and some basic math. While usually unable to live completely independently, many individuals choose to reside in group homes, gain employment, and contribute to their communities in many meaningful ways. Adults with Down syndrome are at increased risk for depression. Increasingly, studies are finding increased risks for Alzheimer's disease, with pathological features evident by age 40 and clinical features by age 50. The expected life span for individuals with Down syndrome is around 30 years, but if deaths during infancy are discounted, the average age of death is closer to 50 years. Survival to 65 years or so is becoming more and more frequent.

Genetics

About 95% of individuals with Down syndrome have trisomy 21: three copies of the 21st chromosome instead of the usual two. This is due to a nondisjunction event during meiosis, and is usually a chance event. Recurrence risks for siblings are generally about 1-2% higher than those for the general population. However, recurrence risks may be higher if either parent carries a translocation or germline mosaicism. Genetic counseling and testing are warranted for parents of every child with Down syndrome.

Management

The diagnosis of Down syndrome is made clinically and confirmed cytogenetically. A chromosomal karyotype should be performed at the time of diagnosis. The diagnosis may be made prenatally.

A formal hearing evaluation should be done in the first year, then annually or biannually after that. The outer ears should be thoroughly cleaned during every evaluation. Ear infections must be treated aggressively. The placement of pressure equalization tubes may be necessary. Amplification options and speech therapy are the primary interventions for hearing loss.

A formal ophthalmologic evaluation should be performed by six months of age, as well as a GI assessment and cardiac assessment including an echocardiogram. Thyroid function testing should be performed in the first year of life, and annually afterwards. Respiratory tract symptoms should be aggressively treated. Due to the possibility of "slippage" of bones in the neck, cervical spine films should be obtained for flexion, neutral, and lateral extension in the first 12-24 months of life.

Following initial evaluations, treatment and management should proceed as needed. A multidisciplinary team is often needed to ensure that all systems are receiving the attention they need. Consultation with a medical geneticist and genetic counselor is indicated. Early interventions such as occupational therapy, physical therapy, and speech therapy should be discussed with the parents.

Resources for Families

Statewide Genetics Program

Phone: 608-267-7148

Fax: 608-267-3824

Email: meyeram@dhfs.state.wi.us

Wisconsin First Step Hotline

Phone: 1-800-642-7837 voice/TTY

Website: www.mch-hotlines.org

Wisconsin Office for Deaf and Hard of Hearing

Phone: 1-608-266-3118 voice/TTY

Website: www.dhfs.state.wi.us/sensory

Regional Children and Youth with Special Health Care Needs Centers

Centers in Green Bay, Wausau, Milwaukee, Madison, and Chippewa Falls

Website: http://dfhs.wisconsin.gov/DPH_BFCH/cshcn/index.HTM

WI Chapter of Families for Hands & Voices

Phone: (920) 437-7370

Website: www.handsandvoices.org

Parent-to-Parent of Wisconsin

Phone: 1-888-266-0028

Email: mathea@shsmh.org

Family Village online resource

Library Card Catalog of Disorders

www.familyvillage.wisc.edu

Down Syndrome WWW Page

<http://nas.com/downsyn/>

National Organization for Rare Disorders (NORD)

www.rarediseases.org