Down Syndrome Facts

Things about the child with Down Syndrome that every parent should know

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Down Syndrome

Down Syndrome is a genetic disorder that occurs in about one out of every 800 births. Down Syndrome is named after Dr John Langdon Down who first identified the syndrome and its moderate mental retardation and physical anomalies. The occurrence of Down Syndrome is not specific to any particular race or socio-economic group.

Down Syndrome is caused by an error in embryonic cell division that replicates an additional chromosome 21 and is called trisomy 21. The cells of the human body contain 23 pairs of chromosomes. At conception each parent supplies 23 chromosomes that pair up after egg fertilization. One half of the 23 chromosomal pairs for the embryonic development of the baby are supplied by the sperm from the male and the other half by the egg from the female. These chromosomes from each parent combine and transmit genetic information that makes the developing human a unique product of the combined genetics of the parents. When a fertilized egg contains an extra chromosome 21 the result is Down Syndrome.

Three Genetic Variations in Down Syndrome

There are three basic genetic variations in Down Syndrome. One variation is called trisomy 21 and occurs 92% of the time. In trisomy 21 the replicated chromosome 21 is present in every cell of the individual. This variation begins in either the sperm or the egg with the presence of the extra chromosome before the egg and sperm unite. The presence of the three copies of the chromosome 21 in this case lead to the name trisomy 21.

The second variation is called mosaic trisomy 21 and occurs in 2%-4% of Down Syndrome. In mosaic trisomy 21 the extra chromosome 21 appears in some but not all cells of the individual. The sperm and egg carry the correct number of chromosomes but there is an error that occurs in the chromosome division of the embryonic development. Only the affected cells with 47 chromosomes instead of the normal 46 will exhibit the Down Syndrome features bringing a partial diversity of Down Syndrome features.

The third variation is called translocation trisomy 21 and occurs in the remaining 3%-4% of Down Syndrome. In this occurrence, either prior to conception or at conception the material from one chromosome 21 becomes translocated or “stuck” to another chromosome 21. The cells of this individual will have 46 chromosomes but still carry features associated with Down Syndrome because of the extra material that is present.

Any of these three variances can contain a "partial trisomy p or q" which means that the child has only a portion of one or the other half of the extra chromosome. The “p” arm of the chromosome being the shorter piece and the “q” arm being the longer piece of the chromosome.

How and When Down Syndrome Occurs

Research has shown that in 88% of Down Syndrome occurrences the replicated chromosome 21 originates from the egg on the maternal side. A smaller 8% of Down Syndrome originates from the sperm of the paternal side. The remaining 2% of occurrences of Down Syndrome are attributed to mitotic or cell division errors that happen after fertilization.

The occurrence of Down Syndrome is medically purported to be a random event in human reproduction. The incidence of Down Syndrome reoccurring in subsequent pregnancies is only about 1% above the baseline maternal age risk unless it is an occurrence of translocation trisomy 21. In translocation trisomy 21 one of the parents may actually be a “balanced carrier” bearing the translocation chromosome. The “balanced

carrier" will have a piece of the 21st chromosome attached to another chromosome (usually the 14th chromosome).

75% of all Down Syndrome occurs to pregnancies where the mother is under the age of 30. However the occurrence of Down Syndrome increases dramatically with the advancement of maternal age. Down Syndrome occurring in a pregnancy with the mother under the age of 30 is less than 1 in 1,000 pregnancies. By the maternal age of 42 Down Syndrome occurs in 1 out of 60 pregnancies. By the maternal age of 49 Down Syndrome occurs in 1 out of 12 pregnancies. (Only 9% of all pregnancies occur in mothers 35 years of age or older).

Prenatal Screening and Diagnostic Testing for Down Syndrome

The simplest prenatal screening test is one that screens the mother's blood for markers for Down syndrome. Lower than normal values for serum alpha feto-protein, low unconjugated estriol and high chorionic gonadotropin values indicate a need for further diagnostic testing to determine with certainty the presence of Down syndrome in the developing fetus. Nuchal Translucency Testing can be done between 11 and 14 weeks of gestation. An ultrasound is used to measure the clear space in the folds of the neck tissue of the fetus. The amount of clear space factored with maternal age is then used to calculate the odds for Down Syndrome and the need for diagnostic testing.

Diagnostic testing for Down syndrome include Percutaneous Umbilical Blood Sampling, Amniocentesis and Chorionic Villous Sampling. Percutaneous Umbilical Blood Sampling is the most accurate diagnostic test but cannot be performed until 18-20 weeks of gestation but carries the greatest risk of miscarriage. Amniocentesis is the analysis of fetal cells taken from the amniotic fluid and carries a much lower risk of miscarriage. This test can be performed at 14 to 18 weeks of gestation. Chorionic Villous Sampling involves sampling a small amount of the tissue near the cervix that develops into the placenta. This procedure can be performed between 9 and 11 weeks of gestation and provides the smallest risk of these tests for miscarriage.

Couples that are at risk of Down Syndrome and other genetic disorders occurring can use a method of in vitro fertilization where the chromosomal balance can be determined before the embryo is implanted in the maternal host.

The Diagnosis of Down Syndrome

Down Syndrome is not always pre-screened and pre-diagnosed. When at birth the baby presents some or all of the features of Down Syndrome tests are ordered to determine any genetic abnormalities. To confirm the presence of the trisomy anomalies a doctor can request a blood test from the baby that grows cells from the blood. Microscopic examination of the cells can then determine the presence or non-presence of the replicated 21st chromosome.

The initial diagnosis of Down Syndrome does not determine the physical or intellectual capabilities or limitations of the child. The developmental challenges in Down Syndrome vary widely and are most affected by the developmental opportunities and interventions afforded to the child.

Many specialized programs and early intervention programs are most helpful to assist in the development of the child with Down Syndrome. The parents of the child with Down Syndrome are encouraged to seek out specialists in the field of Down Syndrome and remain pro-active in the treatments and therapies involved.

Features of Down Syndrome

The features of Down Syndrome can range from mild to severe. Mental and physical development are often slower in the Down Syndrome individual. Intellectual acumen, conceptual and practical skills, social and adaptive behaviors can be limited in the Down Syndrome individual. With these limitations can bring the diagnosis of mild, moderate or severe Mental Retardation. Delayed language development, slow motor skills development and low IQs are often present in Down Syndrome.

Common physical features of Down Syndrome are; Poor muscle tone (hypotonia) and loose ligaments; Small hands and feet with deep creases in the palm of the hand; Short neck, flat face and an upward slant to the eyes (craniofacial dysmorphism); Abnormally shaped ears and white spots in the iris of the eye (Brushfield Spots); skin depigmentation, protruding tongue at birth and limb anomalies.

Medical Disorders Common to Down Syndrome

Common health conditions and medical disorders in Down Syndrome include: Congenital Heart Disease and septal wall defects; bowel and esophagal blockage; Celiac Disease and slow peristalsis; hearing and eye problems including cataracts; skeletal and structural problems; infections and thyroid dysfunction; Dementia and Mental Retardation, seizure disorders and infantile spasms.

Newborn babies with Down Syndrome should be tested for hypothyroidism and congenital heart defects, About half of all children with Down Syndrome have early onset congenital heart disease and pulmonary hypertension (high blood pressure in the lungs).

Due to ear defects in the bones of the inner ear common to Down Syndrome the hearing should be checked periodically. Studies have shown that up to 89% of individuals with Down Syndrome have a significant hearing loss due to both inner ear defects and external ear differences. Amblyopia is common among Down Syndrome and cataracts form in approximately 3% of children with Down Syndrome.

Infants with Down Syndrome are more susceptible to infantile spasms and febrile seizures than other infants. Over 5% of children with Down Syndrome are affected by seizure disorders which is a 10 times greater occurrence for seizure disorders than in the general population.

Help for the Parents of a child with Down Syndrome

Parents of a child diagnosed with Down Syndrome are often overwhelmed at first by feelings of grief, loss, fear and even guilt. Becoming educated about Down Syndrome will help alleviate the fear associated with becoming a new parent of a child with Down Syndrome. Talking with other parents of kids with Down Syndrome can help to deal with the gamut of emotions associated with the initial shock. Consulting with developmental experts can afford great hope and early intervention will establish a pattern of success with the developmental issues associated with Down Syndrome. Developmental pediatricians, speech therapists, occupational therapists and early childhood educators can offer much help and support to accelerate the development of the child with Down Syndrome.

At 3 years of age the child with Down Syndrome is guaranteed educational services under the Individuals with Disabilities Education Act which states that local school districts shall provide "a free appropriate education in the least restrictive environment". An Individualized Education Plan must also be provided for each child. This allows the child with Down Syndrome the opportunity to enjoy the same educational opportunities and activities as other children of the same age.

Each school district has a child study team that will work with you to help you determine the best course for education for your child. The children with Down Syndrome with proactive parents are the children that excel in academic and social skills.

Many children with Down Syndrome develop age appropriate social and academic skills. A few children with Down Syndrome go on to graduate high school with no remedial education. A few go on to college. Many children with Down Syndrome transition to independent or semi-independent living as adults.

Still others continue to live at home but are able to hold jobs, thus finding their own success in the community.

Relationship of Down Syndrome Incidence to Mother's Age

Mother's Age	Incidence of Down Syndrome
Under 30	less than 1 in 1,000
30	1 in 900
35	1 in 400
36	1 in 300
37	1 in 230
38	1 in 180
39	1 in 135
40	1 in 105
42	1 in 60
44	1 in 35
46	1 in 20
48	1 in 16
49	1 in 12