



# Developmental Disabilities

handbook



SANFORD SCHOOL OF MEDICINE

The University of South Dakota

CENTER FOR DISABILITIES

A University Center for Excellence in Developmental Disabilities Education, Research and Service

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This handbook is sponsored in part by a grant from:  
South Dakota Council on Developmental Disabilities

Spring 2003

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# LEGAL DEFINITIONS

## United States Federal Definition - from The Developmental Disabilities Assistance and Bill of Rights Act Amendments of 1994

### Public Law 103-230 [42 USC 6001]

#### (8) Developmental Disability

The term “developmental disability” means a severe, chronic disability of an individual 5 years of age or older that:

- (A) is attributable to a mental or physical impairment or combination of mental and physical impairments;
- (B) is manifested before the individual attains age 22;
- (C) is likely to continue indefinitely;
- (D) results in substantial functional limitations in three or more of the following areas of major life activity:
  - (i) self-care;
  - (ii) receptive and expression language;
  - (iii) learning;
  - (iv) mobility;
  - (v) self-direction;
  - (vi) capacity for independent living;
  - (vii) economic self-sufficiency; and
- (E) reflects the individual’s need for a combination and sequence of special, interdisciplinary, or generic services, supports, or other assistance that is of lifelong or extended duration and is individually planned and coordinated, except that such term, when applied to infants and young children means individuals from birth to age 5, inclusive, who have substantial developmental delay or specific congenital or acquired conditions with a high probability of resulting in developmental disabilities if services are not provided.

## State of South Dakota Definition - from Title 27B, Developmentally Disabled Persons

### 27B-1-18. “Developmental disability” defined.

A developmental disability is any severe, chronic disability of a person that:

- (1) is attributable to a mental or physical impairment or combination of mental and physical impairments;
- (2) is manifested before the person attains age 22;
- (3) is likely to continue indefinitely;
- (4) results in substantial limitations in three or more of the following areas of major life activity; self-care, receptive and expressive language, learning, mobility, self-direction, capacity for independent living, and economic self-sufficiency; and
- (5) reflects the person’s needs for an array of generic services, met through a system of individualized planning and supports over an extended time, including those of a life-long duration.

# ADRENOLEUKODYSTROPHY

**Synonyms** – ALD

**When Detectable** – Varies depending on subtype

**Prevalence** – Estimated 1 in 20,000 to 1 in 50,000

**Ratio** – Varies depending on subtype

## **Description**

First described in 1923, adrenoleukodystrophy was named in 1971 by Dr. Michael Blaw. There are three subtypes of adrenoleukodystrophy.

**Neonatal Adrenoleukodystrophy** (Neonatal ALD) - Both males and females can be affected by Neonatal ALD. This subtype becomes evident after birth. Neonatal ALD is characterized by seizures, delayed psychomotor development, growth deficiencies, severe mental retardation, and possible liver function impairments. Neonatal ALD is usually fatal in infancy or early childhood. Individuals with Neonatal ALD may also exhibit some features of Zellweger Syndrome for that reason it is sometimes classified by itself and not with the other adrenoleukodystrophies - for more information on Zellweger Syndrome, see page 55 of this handbook.

**Childhood Adrenoleukodystrophy** (Childhood ALD) - Only males are affected by Childhood ALD, females may be carriers. Onset is usually between the ages of 4 and 10 years. Characteristics of Childhood ALD are: learning disabilities, perceptual problems, attention deficit disorder (may include hyperactivity), memory loss, impaired vision, personality and behavioral changes, increased skin pigmentation (“bronzing”), nausea and weakness. Childhood ALD is a progressive disorder eventually leading to the inability to communicate, inability to control motor function, dementia and blindness. Childhood ALD is usually fatal within 10 years of the diagnosis.

**Adrenomyeloneuropathy** (AMN) - AMN is a milder form of adrenoleukodystrophy. Adolescent or adult males are affected by AMN and females may be carriers. Characteristics of AMN are: stiffness or clumsiness in the legs, adrenal impairment, general fatigue, urinary difficulties, impotence, nausea and vomiting, and increased skin pigmentation. Cognitive deficits, emotional disturbances, and depressions may also be present. AMN is slowly progressive with its course lasting over several decades with life expectancies being only moderately diminished.

For more information on leukodystrophy, see page 30 of this handbook.

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United Leukodystrophy  
Foundation (ULF)  
2304 Highland Drive  
Sycamore, IL 60178  
Phone 1-800-728-5483  
[www.ulf.org](http://www.ulf.org)

# ANGELMAN SYNDROME

**Synonyms** – AS; Happy Puppet Syndrome (obsolete)

**When Detectable** – Usually diagnosed between the ages of 3 and 7

**Prevalence** – 1 in 15,000 to 1 in 30,000

**Ratio** – 1 male to 1 female

## Description

Angelman Syndrome was first described in 1965 by English physician Dr. Harry Angelman, for whom the syndrome is named.

### Characteristics of Angelman Syndrome

- Developmental delay
- Speech difficulties - diminished or absent speech but with receptive and non-verbal skills
- Unstable gait - often described as “jerky” - walking may be delayed until age 3 or 4 years
- Hand flapping
- Happy demeanor including frequent laughter and smiling
- Mental retardation
- Short attention span
- Possible microcephaly (small head size)
- Possible seizures

Individuals with Angelman Syndrome may benefit from physical therapy. Sign language or picture-based communication boards may be helpful with expressive communication in some individuals with Angelman Syndrome.

It is believed that the incidence of Angelman Syndrome may be higher than currently reported. Angelman Syndrome is sometimes misdiagnosed as cerebral palsy or autism.

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Angelman Syndrome Foundation,  
Inc.  
414 Plaza Drive, Suite 209  
Westmont, IL 60559  
Phone 1-800-IF-ANGEL or  
1-800-432-6435 or  
(630) 734-9267

# APERT SYNDROME

**Synonyms** – ASC I; ASC1; Acrocephalosyndactyly Type 1; Syndactylic Oxycephaly

**When Detectable** – Prenatally with ultrasound or fetoscopy or at birth

**Prevalence** – Estimated at 1 in 160,000 to 1 in 200,000 live births

## Description

Apert Syndrome is named for Dr. Eugene Apert who published a definitive description of the syndrome in 1906.

### Characteristics of Apert Syndrome

- Prematurely fused cranial sutures
- Retruded midface - midface has a sunken-in appearance
- Webbing and/or fusion of the hands and feet including bones and joints of the fingers and toes
- Mental retardation - although normal intelligence has been reported
- Cognitive impairments
- Approximately 30% of individuals with Apert Syndrome have a cleft palate
- Vision problems
- Severe acne
- Recurrent ear infections which can lead to hearing loss

Surgery may be required to remove the pressure placed on the brain by the prematurely fused cranial sutures. Surgery may also be indicated to reshape the retruded midface. Fusion of the fingers and toes may be surgically addressed to provide individuals with Apert Syndrome their highest degree of functionality.

Parents who are not affected by Apert Syndrome and have one child with Apert Syndrome are no more likely to have another child with Apert Syndrome than anyone else in the population. Individuals with Apert Syndrome have a 50% chance of having a child with Apert Syndrome.

Some research indicates increased paternal age as a possible factor in Apert Syndrome.

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AboutFace USA  
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Phone (888) 486-1209  
www.aboutfaceusa.org

Children's Craniofacial Association  
13140 Coit Road  
Suite 307  
Dallas, TX 75240  
Phone 1-800-535-3643 or  
(214) 570-9099  
www.ccakids.com

FACES: The National Craniofacial  
Association  
P.O. Box 11082  
Chattanooga, TN 37401  
Phone 1-800-332-2373  
www.faces-cranio.org



# ARTHROGRYPOSIS

**Synonyms** – Arthrogryposis Multiplex Congenita; AMC

**When Detectable** – At birth

**Prevalence** – Estimated at 1 in 3,000 to 1 in 4,000 live births

**Ratio** – 1 male to 1 female

## Description

First described in 1841 by A.G. Otto who referred to it as “congenital myodystrophy.” The term “arthrogryposis” was first used to describe the condition in 1923.

Arthrogryposis is characterized by multiple joint contractures, meaning there is a limitation in the range of motion in the joint. Arthrogryposis can take a variety of forms. The mildest cases may involve only a few joints with only minimal limitation in the range of motion. The “classic” case of arthrogryposis affects the hands, wrists, elbows, shoulders, hips, feet and knees. Severe cases involve nearly every joint in the body including those in the jaw and back. Muscle weakness may also accompany the joint contractures. Arthrogryposis is non-progressive and has no impact on lifespan.

Two-thirds of all cases of arthrogryposis represent new cases. However, the other one-third of arthrogryposis cases are caused by a hereditary form which is known as “distal arthrogryposis.” Unless the hereditary form - “distal arthrogryposis” - is present parents of one child with arthrogryposis have no greater chance of having another affected child than anyone else in the population.

The non-hereditary cases of arthrogryposis are believed to be caused by something which limits normal joint movement during fetal development. Most sources cite four causes of limitation of fetal joint movement: muscles which do not develop properly, insufficient room in the uterus (which may be the result of low levels of amniotic fluid), central nervous system and spinal cord malformation (these cases are usually accompanied by a wide range of other conditions), and abnormal development of the tendons, bones, joints or joint linings.

Most individuals with arthrogryposis benefit from physical therapy which can improve muscle strength and range of motion in affected joints. In some cases, surgery may be indicated to help other forms of treatment be as beneficial as possible to the individual. Surgery is most often performed on the ankles in order to assist in supporting weight and walking.

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AVENUES: A National Support  
Group for Arthrogryposis  
Multiplex Congenita  
P.O. Box 5192  
Sonora, California 95370  
Phone (209) 928-3668  
www.avenuesforamc.com

# ASPERGER'S SYNDROME

**Synonyms** – AS; Asperger Syndrome; Asperger Disorder; Autism - Asperger's Type

**When Detectable** – Usually not before the age of 3 but before school age

**Prevalence** – Estimated at between 1 in 300 to 2 in 1,000

**Ratio** – Estimated at as high as 7 males to 1 female - all estimates agree males are more often affected than females

## Description

Dr. Hans Asperger first described this syndrome in Germany in 1944. Dr. Asperger's paper was published one year after Dr. Leo Kanner published his paper describing autism.

## Characteristics of Asperger's Syndrome

Nonverbal communication is impaired - may lack facial expressions and use of gestures

Monotonous tone of voice

Tend to speak with single-mindedness about their interests

May fail to understand other's feelings

Poor body awareness which may result in uncoordinated, clumsy or awkward movements

Difficulties in social situations - especially in reactions to social settings

Average or above average intelligence

Highly sensitive to sensory input

Difficulty dealing with changes in environment or schedule

Use and understanding of speech is literal

Individuals with Asperger's Syndrome tend to have specialized interests often focusing on very complex topics such as weather, music, history, geography or astronomy. These interests can often help individuals with Asperger's Syndrome to excel in careers that relate to their specialized interest.

It is not known whether Asperger's Syndrome is a hereditary condition. However, Dr. Asperger's initial research as well as current research indicates a hereditary component as most individuals with Asperger's Syndrome have someone in their family with Asperger-like traits.

Asperger's Syndrome is on the autism spectrum. For more information on the autism spectrum, see page 9 of this handbook.

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Asperger Syndrome Coalition of  
the United States  
PO Box 351268  
Jacksonville, FL 32235-1268  
Phone 1-866-4-ASPRGR or  
1-866-427-7747  
www.asperger.org

Autism Society of America  
7910 Woodmont Avenue  
Suite 300  
Bethesda, MD 20814-3067  
Phone (301) 657-0881 or  
1-800-3-AUTISM or  
1-800-328-8476  
www.autism-society.org

# ATTENTION-DEFICIT/HYPERACTIVITY DISORDER

**Synonyms** – ADHD, ADD, Attention Deficit Disorder

**When Detectable** – Most often diagnosed when the individual begins school

**Prevalence** – Estimated at from 4% to 6% of the population of the United States or from 3% to 5% of school-aged children

**Ratio** – Estimates vary greatly but all estimates agree males are more often affected than females

## **Description**

There are three primary subtypes of Attention-Deficit/Hyperactivity Disorder. The characteristics of the disorder vary depending on subtype.

## **Attention-Deficit/Hyperactivity Disorder**

### **Predominately Inattentive Type**

- Failure to pay close attention
- Making careless mistakes
- Difficulty sustaining attention
- Appears not to listen
- Struggles to follow through on instructions
- Difficulty with organization
- Avoids or dislikes tasks requiring sustained mental effort
- Easily distracted
- May lose things
- Forgetful in daily activities

## **Attention-Deficit/Hyperactivity Disorder**

### **Predominately Hyperactive-Impulsive Type**

- Squirming in chair or fidgeting with hands or feet
- Difficulty remaining seated
- Runs around or climbs on objects excessively
- Difficulty engaging in activities quietly
- Acts as if driven by a motor
- Excessive talking
- Blurting out answers before questions are finished
- Difficulty with waiting or taking turns
- Interrupts or intrudes on others

## **Attention-Deficit/Hyperactivity Disorder**

### **Combined Type**

- Displays characteristics of both Predominately Inattentive Type and Predominately Hyperactive-Impulsive Type.

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Attention Deficit Disorder  
Association (ADDA)  
1788 Second Street, Suite 200  
Highland Park, IL 60035  
www.add.org

Children and Adults with  
Attention-Deficit/Hyperactivity  
Disorder (CHADD)  
8181 Professional Place  
Suite 201  
Landover, MD 20785  
Phone 1-800-233-4050  
www.chadd.org

# AUTISM

**Synonyms** – Infantile Autism; Kanner Syndrome; Autistic Disorder

**When Detectable** – Before the age of 30 months

**Prevalence** – Estimates range from 1 in 5,000 to 1 in 150

**Ratio** – 4 males to 1 female

## Description

Autism was first described in 1943 by American physician Leo Kanner. Dr. Kanner noted that many characteristic behaviors were present in individuals with autism from birth.

### Characteristics of Autism

- Inability to relate to other people
- Failure to make eye contact
- Delayed communication with immature speech rhythms and inappropriate word usage
- Language comprehension is impaired
- Highly sensitive to sensory input - may react indifferently or with an emotional outburst
- Difficulty dealing with changes in environment or schedule
- Inappropriate laughing or giggling may be present
- Stereotypical movements - rhythmic movements may include hand flapping, rocking, spinning or walking on tiptoes
- May develop an inappropriate attachment to an object
- Echolalia (repeating phrases said by others)
- Lack of spontaneous or imaginative play
- Unusual interests that may appear obsessive or compulsive

Intelligence Quotient (IQ) scores vary greatly in individuals with autism. IQ scores range from below 50 to greater than 100. 70% to 75% of individuals with autism have an IQ score of below 70.

The diagnosis of autism is based on observations conducted by trained professionals who evaluate communication, behavior and development. Professionals from many disciplines often work together to diagnose autism.

For more information on autism spectrum disorders, see page 9 of this handbook.

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# AUTISM SPECTRUM DISORDERS

**Synonyms** – Pervasive Developmental Disorders; PDD

**When Detectable** – Varies depending on specific disorder

**Prevalence** – Varies depending on specific disorder

**Ratio** – Varies depending on specific disorder

## Description

Autism Spectrum Disorders refers to five individual and distinct disorders which involve communication and social deficits. These deficits vary in type and severity among the disorders on the spectrum. All of the disorders on the spectrum are classified as pervasive developmental disorders.

This handbook contains specific information for each of the autism spectrum disorders. For more information on a specific disorder, please see the page listed below.

The disorders which make up the autism spectrum are:

Autism - see page 8 of this handbook

Asperger's Syndrome - see page 6 of this handbook

Childhood Disintegrative Disorder - see page 14 of this handbook

Pervasive Developmental Disorder - Not Otherwise Specified -  
see page 44 of this handbook

Rett Syndrome - see page 47 of this handbook

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1-888-777-NAAR or  
1-888-777-6227  
[www.naar.org](http://www.naar.org)

# BECKER MUSCULAR DYSTROPHY

**Synonyms** – BMD

**When Detectable** – In adolescence or adulthood

**Prevalence** – Estimated at 1 in 25,000 males

**Ratio** – Affects males exclusively - females have very rarely been affected but may be carriers

## Description

First described in the 1950's by German physician Peter Emil Becker. This form of muscular dystrophy bears his name. Becker Muscular Dystrophy is a milder form of Duchenne Muscular Dystrophy. All muscular dystrophies result in progressive muscle weakness and wasting as muscle cells are replaced by fat and connective tissue.

## Characteristics of Becker Muscular Dystrophy

- Muscle weakness
- Waddling gait
- Walking on toes
- Walking with stomach extended and shoulders back
- Difficulty climbing stairs
- Cardiac difficulties

Becker Muscular Dystrophy is progressive but its progression is slow, especially when compared to the progression of Duchenne Muscular Dystrophy. Most individuals with Becker Muscular Dystrophy begin using a wheelchair in their thirties while others rely only on canes and other aids well past their thirties.

In 1986, researchers identified the genetic mutation that causes Duchenne Muscular Dystrophy. It is a different mutation on the same gene that causes Becker Muscular Dystrophy. It is important for female relatives of males with Becker Muscular Dystrophy to be tested to determine if they are carriers of this gene.

See also: Duchenne Muscular Dystrophy on page 22 of this handbook.

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Tucson, AZ 85718  
Phone 1-800-572-1717  
www.mdausa.org

Muscular Dystrophy Family  
Foundation  
2330 North Meridian Street  
Indianapolis, IN 46208-5730  
Phone 1-800-544-1213  
www.mdff.org

# BECKWITH-WIEDEMANN SYNDROME

**Synonyms** – BWS; Beckwith Syndrome; Wiedemann-Beckwith Syndrome

**When Detectable** – At birth

**Prevalence** – Estimated at 1 in 15,000 live births

**Ratio** – 1 male to 1 female

## Description

This syndrome was first recognized by Dr. J. Bruce Beckwith working in California in 1963 and by Dr. H.R. Weidemann working in Germany in 1964. The syndrome bears their names.

## Characteristics of Beckwith-Wiedemann Syndrome

Macroglossia (enlarged tongue)

Omphalocele (an abdominal wall defect that allows the intestines to protrude)

Hernia in the area of the navel

Increased growth often expressed as gigantism - stature and weight generally exceed the 90th percentile

Enlarged organs - particularly the kidneys, liver and pancreas

Half of the body may be larger than the other (may not be noticeable until later in life)

Creases and/or pits on the earlobes

Strawberry mark on the forehead or eyelids (sometimes referred to as a “stork bite”) - usually disappears in childhood

Unexplained hypoglycemia (low blood sugar) in the first four months of life - usually disappears within the first year of life

Approximately 5% will develop malignant tumors of the kidneys or adrenal cortex

Mild to moderate mental retardation

Surgery is usually required to repair the omphalocele (abdominal wall defect) shortly after birth. Tongue reduction surgery can be indicated.

It is believed that the prevalence of Beckwith-Wiedemann Syndrome may be greater than currently estimated as mild cases may go undiagnosed.

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Beckwith-Weidemann Support  
Network (BWSN)  
2711 Colony Road  
Ann Arbor, MI 48104  
Phone 1-800-837-2976 or  
(734) 973-0263  
www.beckwith-wiedemann.org

# CEREBRAL PALSY

**Synonyms** – CP; Congenital Cerebral Palsy; Little’s Disease

**When Detectable** – At birth in severe cases, in other cases diagnosis is generally made in early childhood

**Prevalence** – Estimates range from 2 in 1,000 to 4 in 1,000 live births

**Ratio** – 1 male to 1 female

## Description

Cerebral palsy was first described by Dr. William Little in England during the 1860s. It once was known as Little’s Disease. There are three major types of cerebral palsy although some individuals may have characteristics of more than one type.

**Spastic Cerebral Palsy** - This is the most common type of cerebral palsy affecting between 70% and 80 % of the individuals with cerebral palsy. The main characteristic of spastic cerebral palsy is stiff, permanently contracted muscles (which makes movement difficult). Spastic diplegia is used to refer to individuals in whom both legs are affected. Spastic hemiplegia is used to refer to individuals in which one side of the body is affected. Spastic quadriplegia is the most severe form of spastic cerebral palsy affecting all four limbs, the trunk, and the muscles which control the mouth and tongue. Individuals with spastic quadriplegia will also have mental retardation.

**Athetoid or Dyskinetic Cerebral Palsy** - This form affects 10-20% of the individuals with cerebral palsy. The entire body is affected by the dyskinetic form of cerebral palsy. This form is characterized by variations in muscle tone and uncontrolled movements (which range from slow and writhing to rapid and jerky). Muscles of the face and tongue may also be affected causing difficulties with sucking, swallowing and speaking.

**Ataxic Cerebral Palsy** - This form affects 5-10% of the individuals with cerebral palsy. Characteristics of ataxic cerebral palsy include unsteady gait, poor coordination, difficulty with quick or precise movements and depth perception may be affected.

**Mixed Forms** - This term is used to describe individuals with cerebral palsy who display characteristics from more than one form.

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UCP National (United Cerebral  
Palsy)  
1660 L Street, NW  
Suite 700  
Washington, DC 20036  
Phone (202) 776-0406 or  
1-800-872-5827  
www.ucpa.org

March of Dimes Birth Defects  
Foundation  
1275 Mamaroneck Avenue  
White Plains, NY 10605  
Phone 1-888-MODIMES  
or 1-888-663-4637  
www.modimes.org



# CHARCOT-MARIE-TOOTH

**Synonyms** – CMT: Peroneal Muscular Atrophy; Hereditary Motor and Sensory Neuropathy

**When Detectable** – Onset can be in childhood or anytime in adulthood

**Prevalence** – Estimated at 4 in 100,000

**Ratio** – 1 male to 1 female

## Description

First described in 1866 by Dr. Jean M. Charcot and Dr. Pierre Marie in France and Dr. Howard Tooth in England, the syndrome bears the name of all three men. Charcot-Marie-Tooth is the most common inherited neurological disorder.

## Characteristics of Charcot-Marie-Tooth

High arched foot

Degeneration of the nerves below the elbows and below the knees

Muscle atrophy and wasting below the elbows and below the knees

Muscle weakness of the hands, wrists, fingers, feet, ankles and lower legs

Difficulties with balance and walking - may include slapping of the foot as it hits the ground or a high steppage gait

Difficulties with fine motor skills of the hands

Breathing difficulties if the diaphragm is affected

There is a wide variety of severity of the symptoms of Charcot- Marie-Tooth. It is believed that many cases may go undiagnosed because the symptoms are so minor.

Individuals with Charcot-Marie-Tooth do not have a shortened lifespan. Charcot-Marie-Tooth is slowly progressive although some individuals may experience periods of non-progression.

Generally if either parent (mother or father) has Charcot-Marie-Tooth, each child conceived has a 50% chance of developing Charcot-Marie Tooth. However, Charcot-Marie-Tooth can be inherited in several ways and it is often advised that individuals with Charcot-Marie-Tooth receive genetic counseling.

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Charcot-Marie-Tooth Association  
2700 Chestnut Street  
Chester,PA 19013-4867  
Phone 1-800-606-CMTA or  
1-800-606-2682  
www.charcot-marie-tooth.org

# CHILDHOOD DISINTEGRATIVE DISORDER

**Synonyms** – Disintegrative Psychosis; Heller’s Syndrome; Dementia Infantilis

**When Detectable** – After age 2 years and before age 10 years

**Prevalence** – Exact prevalence is unknown but is estimated at 10 times less than autism making it quite rare

**Ratio** – Exact ratio is unknown but boys appear to be more often affected than girls

## Description

First described in 1908 by Austrian special educator Theodore Heller, this disorder sometimes bears his name. Individuals with Childhood Disintegrative Disorder exhibit normal development until onset. The characteristics described below relate to post-onset.

### Characteristics of Childhood Disintegrative Disorder

- Loss of previously acquired social skills
- Loss of bowel and bladder control
- Loss of previously acquired expressive or receptive language
- Loss of previously acquired motor skills
- Loss of previously acquired play skills
- Failure to develop peer relationships
- Nonverbal communication impairment
- Lack of or significant delay in use of spoken language
- Unable to sustain or initiate a conversation with another person

Onset of Childhood Disintegrative Disorder may be very abrupt with loss of skills occurring over a period of days or weeks. However, the onset of Childhood Disintegrative Disorder is more often gradual with the loss of skills occurring over a period of weeks or months.

Just as there are two variations in the rapidity of onset of Childhood Disintegrative Disorder, the disorder can also proceed in a variety of ways. Individuals with Childhood Disintegrative Disorder may experience a progressive deterioration, may achieve a developmental plateau with little improvement or - very infrequently - may achieve a developmental plateau with marked improvement.

For more information on autism spectrum disorders, see page 9 of this handbook.

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Autism and Related Disorders  
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[www.usd.edu/cd/autism](http://www.usd.edu/cd/autism)

Autism Society of America  
7910 Woodmont Avenue  
Suite 300  
Bethesda, MD 20814-3067  
Phone (301) 657-0881 or  
1-800-3-AUTISM or  
1-800-328-8476  
[www.autism-society.org](http://www.autism-society.org)

# CLEFT LIP AND/OR CLEFT PALATE

**Synonyms** – CL/P

**When Detectable** – At birth

**Prevalence** – Estimated at 1 in 700 to 1 in 1,000 live births

Estimated at 1.7 in 1,000 births in Asian populations

Estimated at 3.6 in 1,000 births in certain Native American populations

Estimated at 1 in 2,500 births in African-American populations

**Ratio** – Males are more often affected than females

## Description

**Cleft Lip** (without cleft palate) - a separation of the two sides of the lip - caused by the incomplete closure of the primary palate (the primary palate forms the lip and gums). Cleft lip without cleft palate accounts for about 25% of all cases of clefting.

**Cleft Palate** (without cleft lip) - an opening in the roof of the mouth caused by the incomplete closure of the secondary palate. Cleft palate without cleft lip accounts for about 25% of all cases of clefting.

**Cleft Lip with Cleft Palate** - caused by the incomplete closure of both the primary and secondary palate. Cleft lip with cleft palate accounts for about 50% of all cases of clefting.

Most research indicates that clefts are the result of a combination of genetics and environmental factors.

There is a great range in the severity of clefting. For some individuals, surgery will be required to correct the cleft lip and/or cleft palate. Individuals with cleft lip and/or palate may also have feeding difficulties, fluid in the middle ear, speech difficulties and orthodontic issues. Depending on the associated difficulties, individuals with cleft lip and/or cleft palate may benefit from treatment provided by an interdisciplinary health care team.

It is estimated that 7-13% of individuals with cleft lip without cleft palate are affected by another congenital defect. The percentage rises to 11-14% of individuals with cleft lip and cleft palate who are affected by another congenital defect.

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American Cleft Palate -  
Craniofacial Association /  
Cleft Palate Foundation  
104 South Estes Drive, Suite 204  
Chapel Hill, NC 27514  
Phone (919) 933-9044  
1-800-24-CLEFT or  
1-800-242-5338  
www.cleftline.org

Children's Craniofacial Association  
13140 Coit Road  
Suite 307  
Dallas, TX 75240  
Phone 1-800-535-3643 or  
(214) 570-9099  
www.ccakids.com

# COFFIN-LOWRY SYNDROME

**Synonyms** – CLS; Coffin Syndrome

**When Detectable** – Most often diagnosed in the first years of life

**Prevalence** – Estimated at 1 in 50,000 to 1 in 100,000

**Ratio** – Males are affected more frequently and more severely than females

## Description

First described in 1966 by Dr. G.S. Coffin and in 1971 by Dr. R.B. Lowry, the syndrome is named for both doctors.

### Characteristics of Coffin-Lowry Syndrome

- Short, tapered fingers
- Hands appear puffy with skin that is elastic and loose
- Hypertelorism (wide set eyes)
- Eyes slant downward
- Prominent forehead
- Broad nose
- Large, protruding, low-set ears
- Large mouth with full lips
- Malocclusion (“bad bite”)
- Early loss of baby teeth
- Missing permanent teeth
- Pectus Carinatum (pigeon breast) or Pectus Excavatum (depressed or tunnel chest)
- Kyphosis and/or scoliosis (curvature of the spine)
- Short stature - adult height below the 5th percentile
- Clumsy or ataxic gait
- Mental retardation

Males affected by Coffin-Lowry Syndrome have severe mental retardation.

Females affected by Coffin-Lowry Syndrome have mild mental retardation and are generally affected by a milder form of the syndrome.

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Coffin-Lowry Syndrome  
Foundation (CLSF)  
Attn: Mary C. Hoffman  
3045 255th Avenue S.E.  
Sammamish, WA 98075  
<http://clsfoundation.tripod.com>

# CONGENITAL HYPOTHYROIDISM

**Synonyms** – Hypothyroidism, Congenital; CH; Cretinism

**When Detectable** – At birth

**Prevalence** – Varies depending on type

## Description

There are 3 types of congenital hypothyroidism.

**Thyroid Dysgenesis** - The thyroid gland is either absent, markedly underdeveloped, or not in the correct location in the brain. This type accounts for 80-85% of all cases of congenital hypothyroidism and is estimated to occur in 1 in 4,000 to 1 in 5,000 births.

**Thyroid Dyshormonogenesis** - There is a deficiency or abnormality in the production or release of thyroid hormone. This type accounts for 10-15% of all cases of congenital hypothyroidism and is estimated to occur in 1 in 30,000 births.

**Central Hypothyroidism** - There is a deficiency or abnormality in the production and release of thyroid stimulating hormone (TSH) by the pituitary gland (the pituitary gland along with the hypothalamus is part of the thyroid gland system). This type accounts for less than 5% of all cases of hypothyroidism and is estimated to occur in 1 in 20,000 births.

It is extremely important that all newborns be screened for congenital hypothyroidism. That is why every state in the United States and every province in Canada performs a mandatory screening for congenital hypothyroidism. Screening is performed by a blood test which detects thyroid hormone levels or blood thyroxine.

Babies born with congenital hypothyroidism must be treated with the appropriate dose of thyroid hormone and the thyroid hormone must be given to the child regularly on a daily basis. It is also important for individuals with congenital hypothyroidism to be monitored regularly by a physician.

If congenital hypothyroidism remains undiagnosed and untreated in the first month of life, there will be severe retardation of both brain development and physical growth.

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The Thyroid Society for Education  
and Research  
7515 South Main Street  
Suite 545  
Houston, TX 77030  
Phone (713) 799-9909 or  
1-800-THYROID or  
1-800-849-7643  
[www.the-thyroid-society.org](http://www.the-thyroid-society.org)

# CORNELIA DE LANGE SYNDROME

**Synonyms** – CDLS; de Lange Syndrome; Brachmann-de Lange Syndrome; BDLS

**When Detectable** – At birth

**Prevalence** – Estimated at 1 in 10,000 to 1 in 30,000 live births

**Ratio** – 1 male to 1 female

## Description

This syndrome was first described in 1916 by Dr. W. Brachmann and sometimes bears his name. However, it was Dr. Cornelia de Lange in Holland who first described the complete collection of characteristics of the syndrome and the syndrome is commonly referred to by her name.

## Characteristics of Cornelia de Lange Syndrome

- Low birth weight - usually under five pounds
- Height and weight increase slowly
- Small stature
- Microcephaly (small head size)
- Thin eyebrows that grow together
- Long eyelashes
- Short, upturned nose
- Thin, downturned lips
- Hirsutism (excessive body hair)
- Small hands and feet
- Seizures
- Congenital heart defects
- Cleft palate
- Sucking and swallowing difficulties
- Gastrointestinal difficulties
- Mental retardation ranging from mild to profound
- Developmental delays especially in speech
- Frequent/recurrent respiratory infections

It is not necessary for an individual to exhibit all of these characteristics to be diagnosed with Cornelia de Lange Syndrome.

Cornelia de Lange Syndrome does not shorten the life span. Although some individuals with Cornelia de Lange Syndrome may succumb to infections earlier in life.

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CdLS-USA  
Cornelia de Lange Syndrome  
Foundation Inc.  
302 West Main Street, #100  
Avon, CT 06001  
Phone (860) 676-8166 or  
1-800-223-8355  
[www.cdlsusa.org](http://www.cdlsusa.org)

# CRI DU CHAT SYNDROME

**Synonyms** – Cat Cry Syndrome; 5p- (five p minus) Syndrome; Lejeune Syndrome

**When Detectable** – At birth

**Prevalence** – Estimated at 1 in 20,000 to 1 in 50,000 in the population

**Ratio** – 1 male to 1 female

## Description

First described in 1963 by Dr. Jerome Lejeune in France the syndrome sometimes bears his name. In French, Cri du Chat means cry of the cat which is a term often used to describe the characteristic cry of individuals with this syndrome. This syndrome is also known as 5p- (five p minus) Syndrome. 5p- Syndrome refers to the chromosomal abnormality - a missing portion of chromosome number five- that is present in individuals with the syndrome.

## Characteristics of Cri du Chat Syndrome

- Distinctive, mewing cat-like cry
- Low birthweight
- Microcephaly (unusually small head)
- Poor muscle tone
- Failure to thrive
- Short stature
- Hypertelorism (wide set eyes)
- Eyes may slant downward
- Round face
- Mental retardation ranging from severe to profound
- Language difficulties
- Hair may gray prematurely
- Congenital heart defects in 20% of individuals

Individuals with Cri du Chat Syndrome are often described as friendly and happy. It is noted that individuals with Cri du Chat Syndrome enjoy social interactions.

Cri du Chat Syndrome does not affect the life span.

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5p- Society  
P.O. Box 268  
Lakewood, CA 90714  
Phone (562) 804-4506 or  
(714) 901-1544  
[www.fivepminus.org](http://www.fivepminus.org)

# CYSTIC FIBROSIS

**Synonyms** – CF

**When Detectable** – In infancy

**Prevalence** – Estimated at 1 in 1,600 to 1 in 3,000 live births

**Ratio** – 1 male to 1 female

## Description

Cystic fibrosis was first identified as a specific condition in 1938.

Cystic fibrosis affects the exocrine (outward-secreting) glands causing the body to produce a thick, sticky mucus. This mucus affects the lungs causing respiratory difficulties and the pancreas causing digestive difficulties.

## Characteristics of Cystic Fibrosis

- Very salty tasting skin
- Chronic cough and/or wheezing
- Recurrent pneumonia
- Frequent respiratory infections
- Excessive appetite with poor weight gain
- Bulky, foul-smelling bowel movements

Cystic Fibrosis is among the most common genetic disorders in the Caucasian population. It is estimated that between 1 in 28 and 1 in 30 Americans carry the Cystic Fibrosis gene. When two carriers of the gene conceive a child there is a 25% chance the child will be born with Cystic Fibrosis, a 50% chance the child will be a Cystic Fibrosis carrier, and a 25% chance that the child will not be a carrier. When only one parent is a Cystic Fibrosis carrier, each child has a 50% chance of having Cystic Fibrosis and a 50% chance of being a carrier.

Treatment of Cystic Fibrosis varies with the severity of each case. A specific type of physical therapy uses percussive type movement applied to the chest and/or back of the individual with Cystic Fibrosis can help to loosen the mucus in the lungs. Antibiotics are used to control infections and pancreatic enzymes and nutritional supplements can help to improve digestion.

Females affected by Cystic Fibrosis generally have a poorer prognosis than males affected by Cystic Fibrosis.

Advances in recent years have increased the life expectancy of individuals with Cystic Fibrosis to an average of 30 years (in the 1960's the expected life span was 8 years).

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Boomer Esiason Foundation -  
Fighting Cystic Fibrosis  
200 B Armstrong Road  
Garden City Park, NY 11040  
Phone 516-746-0077  
www.esiason.org

Cystic Fibrosis Foundation  
6931 Arlington Road  
Bethesda, Maryland 20814  
Phone (301) 951-4422 or  
1-800-FIGHTCF or  
1-800-344-4823  
www.cff.org



# DOWN SYNDROME

**Synonyms** – DS; Mosaic 21 Syndrome; Translocation 21 Syndrome; Trisomy 21 Syndrome; Trisomy G Syndrome

**When Detectable** – Prenatally through amniocenteses or chorionic villus sampling or at birth

**Prevalence** – Estimated at 1 in 650 to 1 in 1,000 live births

**Ratio** – 1 male to 1 female

## Description

First described in 1886 by English physician J. Langdon Down, the syndrome bears his name. Down referred to the condition as “Mongolism” a word which is now considered pejorative and obsolete. It wasn’t until 1959 that the chromosomal abnormality responsible for Down Syndrome was identified. The abnormality of chromosome 21 was first identified by Dr. Jerome Lejeune in France. Down Syndrome was the first human condition to be linked to a chromosomal abnormality.

## Characteristics of Down Syndrome

Epicantal folds (vertical folds of skin which hide where the upper and lower eyelids meet on either side of the nose)

Eyes slant slightly upward away from nose

Brushfield spots (speckling on the iris of the eye)

Macroglossia (large tongue that may protrude from mouth)

Short and broad hands

Hypotonia (poor muscle tone)

Mental retardation

Simian crease (single deep crease in the palm of the hand) - present in 30 % of individuals with Down Syndrome

Congenital heart defects - present in 40-50% of individuals with Down Syndrome

Hearing loss - present in 75% of individuals with Down Syndrome

The chances of a women having a child with Down Syndrome increase with her age. Women under the age of 30 have about a 1 in 1,000 chance of having a child with Down Syndrome. However, women over the age of 45 have about a 1 in 32 chance of having a child with Down Syndrome.

Individuals with Down Syndrome are generally described as happy, loving and sociable.

Down Syndrome is the most common identifiable cause of genetic mental retardation and is the most commonly occurring genetic condition.

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National Down Syndrome Society  
666 Broadway  
New York, NY 10012  
Phone (212) 460-9330 or  
1-800-221-4602  
www.ndss.org

Canadian Down Syndrome Society  
811-14 Street N.W.  
Calgary, Alberta,  
Canada T2N 2A4  
Phone (403) 270-8500  
www.cdss.ca

# DUCHENNE MUSCULAR DYSTROPHY

**Synonyms** – DMD; Psuedohypertrophic Muscular Dystrophy

**When Detectable** – In early childhood, usually between the ages of 2 and 6 years

**Prevalence** – Estimated between 1 in 3,000 and 1 in 4,000 male births

**Ratio** – Affects males exclusively - females have very rarely been affected but may be carriers

## Description

First described in 1861 by Dr. Guillaume B.A. Duchenne in France, this type of muscular dystrophy bears his name. Duchenne Muscular Dystrophy is the most common and severe form of muscular dystrophy. All muscular dystrophies result in progressive muscle weakness and wasting as muscle cells are replaced by fat and connective tissue.

## Characteristics of Duchenne Muscular Dystrophy

Muscle weakness as early as 3 years of age

Delayed walking

Enlarged calf muscles

By school age - walking on the toes or balls of the foot, stumbling, difficulty climbing stairs, unsteady gait, may fall easily

May walk with stomach extended and shoulders back to improve gait

Between 7 and 12 years of age, loss of the ability to walk

Skeletal contractures

Muscle atrophy

Breathing becomes difficult in the final stages

Of the individuals with Duchenne Muscular Dystrophy, approximately one-third will be affected by mental retardation.

Most individuals with Duchenne Muscular Dystrophy survive into their twenties. Duchenne Muscular Dystrophy results in fatal respiratory or cardiac failure.

In 1986, researchers identified the genetic mutation that causes Duchenne Muscular Dystrophy. It is important for female relatives of males with Duchenne Muscular Dystrophy to be tested to determine if they are carriers of this gene.

See also: Becker Muscular Dystrophy on page 10 of this handbook.

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Muscular Dystrophy Association  
(MDA) - USA  
National Headquarters  
3300 East Sunrise Drive  
Tucson, AZ 85718  
Phone 1-800-572-1717  
www.mdaua.org

Muscular Dystrophy Family  
Foundation  
2330 North Meridian Street  
Indianapolis, IN 46208-5730  
Phone 1-800-544-1213  
www.mdff.org

# EPILEPSY

**When Detectable** – Onset can occur anytime

**Prevalence** – Between 1% and 2% of the general population

**Ration** - 1 male to 1 female

## Description

Epilepsy is a neurological condition that produces disturbances in the brain's normal function. These disturbances produce seizures.

There are three types of epilepsy.

**Idiopathic Epilepsy** - In this type there is no known cause of the epilepsy. Seven out of 10 individuals with epilepsy have this type.

**Symptomatic Epilepsy** - This type is usually the result of a structural abnormality in the brain that is either present at birth or occurring later in life.

**Cryptogenic Epilepsy** - In this type there is no known cause of the epilepsy but physicians strongly suspect a cause.

There are five basic forms of seizures that affect individuals with epilepsy.

**Simple Partial Seizures** - Characteristics are strange or unusual sensation, sudden restless movement, hearing or vision distortion, stomach discomfort, sense of fear, and consciousness is not impaired.

**Complex Partial Seizures** - Characteristics are confusion with complicated motor action. Individual loses awareness (can't remember their actions) but doesn't lose consciousness. This type of seizures affect an estimated two-thirds of all individuals with epilepsy.

**Generalized Absence Seizures** - This type of seizure generally affects children. Individual loses awareness and may stare into space followed by a return to normal activity.

**Tonic-Clonic Seizures** - This type of seizure involves two phases. In the first or tonic phase, the individual loses consciousness, their body becomes rigid and they fall. In the second or clonic phase, the individual's body jerks and twitches. Consciousness returns slowly.

**Status Epilepticus** - This type describes a state in which an individual has a series of recurring seizures between which consciousness does not return. This type of seizure requires immediate medical attention.

Epilepsy has been known since ancient times when it was often thought that individuals with epilepsy had been touched by the gods. In fact the word epilepsy is derived from the Greek word "epilepsia" meaning to take hold of or seize.

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www.usd.edu/cd

Wegner Health Science  
Information Center  
1400 West 22nd Street  
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Sioux Falls, SD 57105  
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Epilepsy Foundation of America  
4351 Garden City Drive  
Landover, MD 20785-7223  
Phone 1-800-332-1000  
www.efa.org

Epilepsy Canada  
National Office  
1470 Peel Street  
Suite 745  
Montreal Quebec H3A 1T1  
Phone 1-877-SEIZURE or  
1-877-734-9873  
www.epilepsy.ca

# FETAL ALCOHOL SPECTRUM DISORDERS

**Synonyms** – Varies depending on type

**When Detectable** – At birth in severe cases or throughout the lifespan

**Prevalence** – Across the spectrum estimated at 1% of all births or between 20 in 10,000 and 100 in 10,000 births

**Ratio** – 1 male to 1 female

## Description

Fetal Alcohol Spectrum Disorders (FASD) refers to the entire spectrum of physical, mental and behavioral abnormalities caused by the consumption of alcohol by women while they are pregnant. It is the leading cause of mental retardation. It is **100% Preventable**.

The most recent diagnostic criteria divides FASD into five distinct categories.

- 1 - **Fetal Alcohol Syndrome (FAS) with Confirmed Maternal Alcohol Exposure**
- 2 - **Fetal Alcohol Syndrome (FAS) without Confirmed Maternal Alcohol Exposure**
- 3 - **Partial Fetal Alcohol Syndrome (PFAS) with Confirmed Maternal Alcohol Exposure**
- 4 - **Alcohol-Related Birth Defects (ARBD)**
- 5 - **Alcohol-Related Neurodevelopmental Disorder (ARND)**

Characteristics of Fetal Alcohol Spectrum Disorder fall into three major categories.

- Growth Deficiency** - in height, in weight, in both height and weight, either prenatally or postnatally
- Facial Anomalies** - may include: infantile philtrum (ridges between the nose and mouth), short palpebral fissures (eye slits), flat midface, short upturned nose, thin upper lip
- Central Nervous System Dysfunction** - may include: microcephaly (small brain size), seizures, hyperactivity, fine and/or gross motor difficulties, attention deficits, learning disabilities, mental retardation, developmental delays, intellectual disabilities

For more information on Fetal Alcohol Spectrum Disorder, please see the Center for Disabilities' *Fetal Alcohol Syndrome Handbook*. The handbook is available online at [www.usd.edu/cd/fashandbook](http://www.usd.edu/cd/fashandbook) or by contacting the Center for Disabilities.

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National Organization on Fetal  
Alcohol Syndrome (NOFAS)  
216 G Street, North East  
Washington, DC 20002  
Phone (202) 785-4585  
[www.nofas.org](http://www.nofas.org)

Fetal Alcohol Syndrome Family  
Resource Institute (FAS\*FRI)  
PO Box 2525  
Lynnwood, WA 98036  
Phone (253) 531-2878  
[www.fetalalcoholsyndrome.org](http://www.fetalalcoholsyndrome.org)

March of Dimes Birth Defects  
Foundation  
1275 Mamaroneck Avenue  
White Plains, NY 10605  
Phone -1-888-MODIMES or  
1-888-663-4637  
[www.modimes.org](http://www.modimes.org)

# FRAGILE X SYNDROME

**Synonyms** – FRAXA, Martin-Bell Syndrome, Marker X Syndrome, Fragile X Mental Retardation

**When Detectable** – Prenatally through amniocentesis, diagnosis may be made in childhood based on intellectual delays or at puberty based on physical characteristics

**Prevalence** – Estimated at 1 in 2,000 to 1 in 3,600 male births  
Estimated at 1 in 4,000 to 1 in 6,000 female births

**Ratio** – Estimated at 2 males to 1 female

## Description

Fragile X Syndrome was first identified by geneticist Herbert Lubs in 1969. Fragile X Syndrome derives its name from the fact that individuals with the syndrome have a pronounced gap on the long arm of the X chromosome. This gap tends to break or tear when studied in a laboratory - in other words it is "fragile." Therefore the syndrome was named Fragile X.

## Characteristics of Fragile X Syndrome

Intellectual disability - may range from a mild learning disability to severe mental retardation

Enlarged ears

Long face with prominent chin

Macroorchidism (large testicles)

Double jointed - especially in the fingers

Macrosomia (large body size) in childhood

Autistic-like behaviors may include hand-flapping, failure to make eye contact, and aversion to touch and noise

Speech difficulties

Anxiety

Females tend to be more mildly affected by Fragile X Syndrome.

Approximately one-third of females with Fragile X Syndrome have a significant intellectual disability.

Fragile X Syndrome is the second most common identifiable cause of genetic mental retardation. Down Syndrome is the most common. It is estimated that 5% to 10 % of all males with mental retardation have Fragile X Syndrome.

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Conquer Fragile X Foundation  
450 Royal Palm Way  
Suite 400  
Palm Beach, FL 33480  
Phone (561) 842-9219  
www.cfxf.org

FRAXA Research Foundation  
45 Pleasant Street  
Newburyport, MA 01950  
Phone (978) 462-1866  
www.fraxa.org

National Fragile X Foundation  
P.O. Box 190488  
San Francisco, CA 94119  
Phone 1-800-688-8765  
www.nfxf.org

# GOLDENHAR SYNDROME

**Synonyms** – Goldenhar-Gorlin Syndrome; hemifacial microsomia; oculoauriculo-vertebral spectrum; oculoauricular dysplasia (OAV)

**When Detectable** – Prenatally with ultrasound or at birth

**Prevalence** – Estimated at 1 in 3,000 to 1 in 5,000

**Ratio** – 3 males to 2 females

## Description

First described in 1952 by French physician M. Goldenhar for whom the syndrome is named.

### Characteristics of Goldenhar Syndrome

- Microtia (a partial formed or totally absent ear)
- Under development of the mandible
- Hemifacial Microsomia (one side of the face is smaller than the other)
- Missing eyes
- One corner of the mouth may be higher than the other
- Benign growths in the eyes
- Macrosomia (a lateral cleft which makes the mouth appear larger)
- Cervical Hemivertebrae (vertebrae which are small or not completely formed on one side)
- Moderate learning disabilities in 10-15% of affected individuals

Individuals with Goldenhar Syndrome may experience hearing problems ranging from mild to severe which are generally due to the structure of the ear canal or of the bones involved in hearing. Vision problems are also frequently present in individuals with Goldenhar Syndrome.

Dental problems may require surgery to correct mandibular retrusion. Cosmetic surgery may be used to lessen the appearance of facial asymmetry.

Goldenhar Syndrome does not affect lifespan.

Parents of a child with Goldenhar Syndrome have a 1% to 2% chance of having another child affected by the syndrome. Individuals with Goldenhar Syndrome have a 3% chance of having a child affected by the syndrome.

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FACES: The National Craniofacial  
Association  
P.O. Box 11082  
Chattanooga, TN 37401  
Phone 1-800-332-2373  
www.faces-cranio.org

World Craniofacial Foundation  
7777 Forest Lane, Suite C-621  
P.O. Box 515838  
Dallas, TX 75251-5838  
Phone (972) 566-6669 or  
1-800-533-3315  
www.worldcf.org

# KRABBE DISEASE

**Synonyms** – Globoid Cell Leukodystrophy; GCL

**When Detectable** – Usually between the ages of 3 and 6 months

**Prevalence** – Estimated at 1 in 100,000 births in the United States

**Ratio** – 1 male to 1 female

## Description

First described by Dr. K.H. Krabbe in Demark in 1916, the disease is named for him. Krabbe Disease is a form of leukodystrophy.

The progression of Krabbe Disease has been divided into 3 stages.

### Stage 1

- Irritability
- Stiffness of the limbs
- Mental and motor development is stopped
- Fever without infection

### Stage 2

- Severe arching of the back
- Deterioration of learned skills
- Periods of hypertonia (increased muscle tone)
- Fever

### Stage 3

- Debilitation with no voluntary movement

Individuals with Krabbe Disease usually succumb to respiratory infection or cerebral hyperpyrexia by age 1 year though some have been known to live until their third or fourth year.

The information on this page deals specifically with the infantile form of Krabbe Disease. The infantile form affects 90% of the individuals with Krabbe Disease. There are also late infantile, juvenile, adolescent and adult versions of this disease. The characteristics of each of these types varies greatly as does the life expectancy.

For more information on leukodystrophy, see page 30 of this handbook.

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Hunter's Hope Foundation  
PO Box 643  
Orchard Park, NY 14127  
Phone (716) 667-1200 or  
1-877-984-HOPE or  
1-877-984-4673  
www.huntershope.org

United Leukodystrophy  
Foundation (ULF)  
2304 Highland Drive  
Sycamore, IL 60178  
Phone 1-800-728-5483  
www.ulf.org

# KLINEFELTER SYNDROME

**Synonyms** – Klinefelter’s Syndrome; 47 XXY; 48 XXYY; 48 XXXY; 49 XXXXY, XY/XXY Mosaic

**When Detectable** – Various times across the lifespan

**Prevalence** – Estimated at 1 in 500 to 1 in 1,000 live male births

**Ratio** – Specific to males

## Description

First described in 1942 by American Dr. H.F. Klinefelter, the syndrome bears his name. All men have one X chromosome and one Y chromosome.

Klinefelter Syndrome, in most cases, is caused by an extra X chromosome.

## Characteristics of Klinefelter Syndrome

Hypogonadism (small testes, small penis, inadequate testosterone - the male hormone - production)

Infertility

Tall stature - legs may not be proportional to the arms and trunk

Below average scores on intelligence tests by 20% of individuals with Klinefelter Syndrome

Learning disabilities

Sparse or thin body hair

Gynecomastia (enlarged breasts) present in 50% of individuals with Klinefelter Syndrome

Shyness

Social difficulties

Speech and language difficulties may be present

Psychological problems may be present including lack of self-esteem, anxiety and depression

Dental problems may be present

Increased risk for cancer of the breast tissue

There are four times when the diagnosis of Klinefelter Syndrome may be made.

**Prenatally** - through amniocentesis

**In Childhood** - when developmental delays are present

**At Puberty** - when physical development is not typical

**In Adulthood** - during fertility studies

It is estimated that between 1 in 25 and 1 in 75 men seeking infertility treatment have Klinefelter Syndrome.

Klinefelter Syndrome does not affect the lifespan

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American Association for  
Klinefelter Syndrome Information  
and Support (AAKSIS)  
2945 W. Farwell Avenue  
Chicago, IL 60645-2925  
Phone 1-888-466-KSIS or  
1-888-466-5747  
www.aaksis.org

Klinefelter Syndrome and  
Associates  
P.O. Box 119  
Roseville, CA 95678-0119  
Phone 1-888-XXY-WHAT or  
1-888-999-9428  
www.genetic.org/ks



# LEARNING DISABILITIES

## Synonyms – LD

**When Detectable** – At anytime - often diagnosed when an individual begins school

**Prevalence** – Estimate range from 10% to 15% of the general population

## Description

Learning disabilities is an “umbrella” term used to cover a wide variety of different learning difficulties. In general, learning disabilities affect an individual’s ability to process, store and express information. They may be expressed as difficulties thinking, listening, speaking, writing, spelling or doing mathematical calculations.

Some common types of learning disabilities are:

**Dyslexia** - a difficulty with language processing expressed by difficulty reading, writing and spelling

**Dysgraphia** - a difficulty with the motor patterns used in writing

**Dyscalculia** - a difficulty with math skills

## Some Common Signs of Learning Disabilities

Short attention span

Difficulty recognizing mistakes

Difficulty remembering information

Difficulty recalling instructions

Using words inappropriately when speaking

Difficulty expressing thoughts concisely

Difficulty reading - may be either actual difficulty reading (reversing letters, etc.) or difficulty with comprehension

Difficulty writing - may include reversing letters or words

Poor handwriting

Difficulty spelling

Difficulty with written mathematical calculations

Difficulty calculating money

Learning disabilities are NOT the same as mental retardation, autism, sensory impairment, behavioral disorders or Attention Deficit/Hyperactivity Disorder.

Learning disabilities are NOT caused by environmental factors such as economic disadvantages, cultural differences, or insufficient/inappropriate education.

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National Center for Learning  
Disabilities  
381 Park Avenue  
South Suite 1401  
New York, NY 10016  
Phone (212) 545-7510 or  
1-888-575-7373  
www.nclد.org

Council for Learning Disabilities  
P.O. Box 40303  
Overland Park, KS 66204  
Phone (913) 492-8755  
www.cldinternational.org

Learning Disabilities Association of  
America  
4156 Library Road  
Pittsburgh, PA 15234-1349  
Phone (412) 341-1515  
www.ldanatل.org

# LEUKODYSTROPHY

**Synonyms** – White Matter Disease; others depending on type

**When Detectable** – Varies depending on type

**Prevalence** – Varies depending on type

**Ratio** – Varies depending on type

## Description

Leukodystrophy is a term applied to a group of progressive, degenerative, genetic diseases. It takes its name from the Greek *leuko* (white), *dys* (disordered), and *trophy* (growth). *Leuko* or white is used because leukodystrophies affect the white matter of the central nervous system. The white matter appears white because it contains a complex chemical substance called myelin. Myelin forms a sheath around the axon which is a strand of nerve fiber that conducts nerve impulses. In individuals affected by a leukodystrophy the myelin sheath is attacked and often destroyed. There are several distinct leukodystrophies because each affects one - and only one - of the many distinct chemicals that make up the myelin sheath.

The following are the types of leukodystrophies. Those covered more extensively elsewhere in this handbook are noted.

Adrenoleukodystrophy - See page 2 of this handbook.

Aicardi-Goutiers Syndrome

Alexander(s) Disease

CACH

CADASIL

Canavan Disease

Cerebrotendinous Xanthomatosis (CXT)

Krabbe Disease - See page XX of this handbook.

Metachromatic Leukodystrophy - See page 32 of this handbook.

Neonatal Adrenoleukodystrophy - See page 2 of this handbook.

Ovarioleukodystrophy Syndrome

Pelizaeus-Merzbacher Disease

Refsum Disease

Van der Knaap Syndrome

Zellweger Syndrome - See page 55 of this handbook.

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United Leukodystrophy  
Foundation (ULF)  
2304 Highland Drive  
Sycamore, IL 60178  
Phone 1-800-728-5483  
www.ulf.org

# MARFAN SYNDROME

**Synonyms** – Arachnodactyly, Contractural Arachnodactyly, Marfanoid Hypermobility Syndrome

**When Detectable** – At birth if suspected or diagnosis can be made at anytime during the lifespan

**Prevalence** – Estimated from 1 in 10,000 to 3 in 200,000

**Ratio** – 1 male to 1 female

## Description

First described in 1896 by the French physician Bernard-Jean Antonin Marfan for whom the syndrome is named.

### Characteristics of Marfan Syndrome

- Mitral valve prolapse which can lead to irregular heartbeat
- Aorta (the main artery carrying blood away from the heart) may be wider and more fragile
- Scoliosis (curvature of the spine)
- Pectus deformity (abnormally shaped chest) may be expressed as either pectus carinatum (pigeon breast or protruding breastbone) or pectus excavatum (funnel chest or indented breast bone)
- Hypermobility (joints that bend beyond the usual limit)
- Dolichostenomelia (disproportionally long arms and legs)
- Arachnodactyly (long, thin fingers)
- Tall stature may be present
- Mypopia (nearsightedness)
- Dislocation of the ocular lens present in 50% of individuals with Marfan Syndrome

The most common cause of death for individuals with Marfan Syndrome is complications related to cardiac abnormalities. Individuals with few or mild physical characteristics of Marfan Syndrome may only be diagnosed after their death due to cardiac complications.

Each child born to an individual with Marfan Syndrome has a 50% chance of inheriting the syndrome.

Historians speculate, based on physical characteristics, that individuals such as the violinist Niccolo Paganini, the composer Rachmaninoff, Mary Queen of Scots and possibly the Egyptian Pharaoh Akhenaten were affected by Marfan Syndrome. However, as there is no specific laboratory test used to diagnose Marfan Syndrome this is only speculation.

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National Marfan Foundation  
(NMF)  
382 Main Street  
Port Washington, NY 11050  
Phone (516) 883-8712 or  
1-800-8-MARFAN or  
1-800-862-7326  
www.marfan.org

American Heart Association  
7272 Greenville Avenue  
Dallas, TX 75231  
Phone 1-800-AHA-USA-1 or  
1-800-242-8721  
www.americanheart.org

Canadian Marfan Association  
Centre Plaza Postal Outlet  
128 Queen Street South  
P.O. Box 42257  
Mississauga, Ontario, L5M 4Z0,  
Canada  
Phone (905) 826-3223  
www.marfan.ca

# METACHROMATIC LEUKODYSTROPHY

**Synonyms** – MLD

**When Detectable** – Varies depending on type

**Prevalence** – Estimated at 1 in 40,000 to 1 in 50,000 live births

**Ratio** – 1 male to 1 female

## **Description**

There are 3 types of metachromatic leukodystrophy (MLD).

**Late Infantile MLD** - Usually symptoms appear between 6 months and 2 years of age. Growth is normal until onset when skills (walking, talking and others) begin to deteriorate. Progression is rapid with the child eventually losing voluntary muscle control, becoming blind, unable to communicate and having difficulty swallowing. Late infantile MLD is usually fatal between the ages of 3 and 6 years. This is the most common type of MLD.

**Juvenile MLD** - Most often diagnosed during the early school years (between 4 and 12 years of age) when scholastic performance of the individual begins to fall. Incontinence, difficulty walking and slurred speech may also be present. Progression may bring seizures, tremors, abnormal postures and possibly the inability to walk. The final stages of juvenile MLD are similar to those of late infantile MLD. Life expectancy has increased in recent years with some individuals affected by juvenile MLD living into adulthood.

**Adult MLD** - Onset may occur anytime beginning at 14 years of age and has been described as late as sometime in the 60s. Characteristics that appear at onset are a change in personality, emotional changes, and decreased job performance. Adult MLD is sometimes misdiagnosed as schizophrenia or depression. Progressive loss of cognitive and motor functions generally occurs over 10 to 30 years.

For more on leukodystrophy, see page 30 of this handbook.

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2304 Highland Drive  
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Phone 1-800-728-5483  
www.ulf.org

# MENTAL RETARDATION

**Synonyms** – MR

**When Detectable** – Depending on cause at birth or in childhood

**Prevalence** – Estimated at between 2.5 % and 3% of the general population or an estimated 6.2 to 7.5 million Americans

**Ratio** – Estimated to be more common in males than females

## Description

Mental retardation is a developmental disability that is defined as a limitation in intellectual functioning and in adaptive behavior that begins before the age of 18 years. An Intelligence Quotient (IQ) score of below 75, while no longer necessary for a diagnosis of mental retardation, can still be used as a representation of an intellectual limitation.

Some examples of adaptive behavior (those skills in which a person must be limited for a diagnosis of mental retardation) are:

**Conceptual Skills** - receptive language, expressive language, reading, writing, money concepts, self-direction

**Social Skills** - responsibility, self-esteem, following rules, avoiding victimization, interpersonal skills

**Practical Skills** - activities of daily living, using the instruments of daily living (telephone, transportation, money, etc.), occupational skills

In about one-third of the individuals with mental retardation, the cause is unknown. The known causes of mental retardation can be broken into five broad categories.

**Genetic Conditions** - for example - untreated Phenylketonuria, Fragile X Syndrome, Down Syndrome

**Problems During Pregnancy** - for example - maternal alcohol use, maternal drug use, maternal cytomegalovirus, maternal rubella

**Problems at Birth** - for example - injury to the infant's brain, prematurity

**Problems After Birth** - for example - accidents (such as a blow to the head or near drowning), lead poisoning, meningitis

**Social Factors** - for example - malnutrition, inadequate medical care, environmental health hazards, under-stimulation

Some causes of mental retardation can be prevented (such as Fetal Alcohol Syndrome and untreated Phenylketonuria) while others (such as Down Syndrome and Fragile X Syndrome) cannot be prevented.

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American Association on Mental  
Retardation  
444 North Capital Street  
Washington, DC 20001-1512  
Phone (202) 387-1968  
www.aamr.org

The Arc  
1010 Wayne Avenue  
Suite 650  
Silver Spring, MD 20910  
Phone (301) 565-3842  
www.thearc.org

# MUCOPOLYSACCHARIDOSIS

## Synonyms – MPS

**When Detectable** – In early childhood or prenatally through enzyme assay of the chorionic villi or amniotic fluid

**Prevalence** – Varies depending on type but across all types estimated at 1 in 25,000 live births

**Ratio** – 1 male to 1 female - except Type II, Hunter Syndrome, which is exclusive to males

## Description

There are six recognized types of mucopolysaccharidosis (MPS) each type may encompass several sub-types. Each type is distinct but all are the result of a deficiency of a certain enzyme which is necessary for the body to metabolize mucopolysaccharides (a complex carbohydrate) causing the body to store the mucopolysaccharides in its cells.

**MPS I** Hurler Syndrome  
Scheie Syndrome  
Hurler-Scheie Syndrome

**MPS II** Hunter Syndrome, mild  
Hunter Syndrome, severe

**MPS III** San Filippo A  
San Filippo B  
San Filippo C  
San Filippo D

**MPS IV** Morquio A  
Morquio B

**MPS V** Vacant - formerly Scheie Syndrome which has been reclassified as MPS I

**MPS VI** Maroteaux-Lamy, classic severe  
Maroteaux-Lamy, intermediate  
Maroteaux-Lamy, mild

**MPS VII** Sly Syndrome

For more information on the mucopolysaccharidosis types or subtypes, please see pages 35-40 of this handbook.

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102 Aspen Drive  
Downington, PA 19335  
Phone (610) 942-0100  
www.mpssociety.org

The Canadian Society for  
Mucopolysaccharide & Related  
Diseases, Inc.  
P.O. Box 64714  
Union Ville, Ontario  
L3R 0M9 CANADA  
Phone (905) 479-8701 or  
1-800-667-1846  
www.mpssociety.ca

National Tay-Sachs & Allied  
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2001 Beacon Street  
Suite 204  
Brighton, MA 02135  
Phone 1-800-906-8723  
www.ntsad.org

# MUCOPOLYSACCHARIDOSIS I

## Synonyms - MPS I

**When Detectable** – In early childhood or prenatally through enzyme assay of the chorionic villi or amniotic fluid

**Prevalence** – Varies depending on sub-type, see below

**Ratio** – 1 male to 1 female

## Description

There are 3 sub-types of mucopolysaccharidosis I (MPS I).

**Hurler Syndrome (MPS IH)** - First described in 1919 by Dr. Gertrud Hurler in Germany. Hurler Syndrome is the most severe of the MPS I sub-types. Hurler Syndrome is usually fatal by age 10 years. Prevalence is estimated at 1 in 100,000. Characteristics of Hurler Syndrome are:

- Growth deficiency - with growth often stopping by age 2
- Coarse facial features - prominent forehead, thick earlobes, full lips, and low nasal bridge
- Stiff joints
- Cloudy corneas
- Severe mental retardation
- Hepatosplenomegaly (enlargement of the spleen and liver)

**Scheie Syndrome (MPS IS)** - First described in 1962 by ophthalmologist Dr. Harold Scheie at the University of Pennsylvania. Prevalence is estimated at 1 in 500,000. Characteristics of Scheie Syndrome are:

- Moderately short stature
- Joint contractures (limitation of the range of motion in the joint)
- Cloudy corneas - may lead to blindness
- Normal intelligence
- Possible psychotic episodes

**Hurler-Scheie Syndrome (MPS IH/S)** - A compound of Hurler and Scheie Syndromes, Hurler-Scheie Syndrome is usually the diagnosis when there is normal or near normal intelligence but with more severe physical manifestations than Scheie Syndrome. Prevalence is estimated at 1 in 115,000.

For more information on mucopolysaccharidosis (MPS), please see page 34 of this handbook.

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# MUCOPOLYSACCHARIDOSIS II

**Synonyms** – MPS II; Hunter Syndrome; gargoylism (obsolete)

**When Detectable** – In early childhood or prenatally through enzyme assay of the chorionic villi or amniotic fluid

**Prevalence** – Estimated at 1 in 100,000 to 1 in 150,000 births

**Ratio** – Affects males exclusively - females have very rarely been affected but may be carriers

## Description

There are two sub-types of mucopolysaccharidosis II (MPS II). Both are named for Dr. Charles Hunter who in 1917 first described them in Canada.

**Hunter Syndrome, mild** - The characteristics of Hunter Syndrome, mild are:

- Growth deficiency
- Coarse facial features - flat nose with wide nostrils and depressed nasal bridge, thick lips and tongue
- Mental function may deteriorate slowly and progressively after age 5 or 6 years
- Stiffening of joints
- Hypertrichosis (excessive hair)
- Hepatosplenomegaly (enlargement of the spleen and liver)

**Hunter Syndrome, severe** - The characteristics of Hunter Syndrome, severe are:

- Growth deficiency
- Coarse facial features - flat nose with wide nostrils and depressed nasal bridge, thick lips and tongue
- Severe mental retardation
- Stiffening of joints
- Hypertrichosis (excessive hair)
- Hepatosplenomegaly (enlargement of the spleen and liver)
- Respiratory infections
- Cardiac complications

Individuals with Hunter Syndrome, mild have been known to live into their 60s. Individuals with Hunter Syndrome, severe typically live between 5 and 14 years. Sisters and maternal aunts of an individual with Hunter Syndrome may be carriers and are urged to undergo genetic testing.

For more information on mucopolysaccharidosis (MPS), please see page 34 of this handbook.

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# MUCOPOLYSACCHARIDOSIS III

**Synonyms** – MPS III, Sanfilippo Syndrome

**When Detectable** – In early childhood or prenatally through enzyme assay of the chorionic villi or amniotic fluid

**Prevalence** – Estimated at 1 in 24,000 births

**Ratio** – 1 male to 1 female

## Description

There are four sub-types of mucopolysaccharidosis III (MPS III). The sub-types are Sanfilippo Syndrome A, Sanfilippo Syndrome B, Sanfilippo Syndrome C, and Sanfilippo Syndrome D. Each sub-type is the result of the deficiency of a certain and unique enzyme. However, there is very little difference in the manifestation of the four sub-types so they will be treated as one here.

MPS III was first described by Dr. Sylvester J. Sanfilippo in 1963.

## Characteristics of MPS III

Normal or accelerated growth in the first 3 years of life followed by slow growth

Mildly restricted movement of the knees and elbow

Severe mental retardation

Restlessness

Overactivity

Aggressiveness

Behavioral problems

Diminished attention span

Sleep disturbances - resulting in very little sleeping at night

Individuals with Sanfilippo Syndrome (MPSIII) eventually lose their ability to use language and their ability to walk. Affected individuals may eventually enter a vegetative state. Most individuals with Sanfilippo Syndrome live into adolescence however about one-third of affected individuals may survive into adulthood.

For more information on mucopolysaccharidosis (MPS), please see page 34 of this handbook.

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# MUCOPOLYSACCHARIDOSIS IV

**Synonyms** – MPS IV; Morquio Syndrome, Morquio-Brailsford Syndrome

**When Detectable** – In early childhood or prenatally through enzyme assay of the chorionic villi or amniotic fluid

**Prevalence** – Estimated at 1 in 40,000 to 1 in 200,000 births

**Ratio** – 1 male to 1 female

## Description

Mucopolysaccharidosis IV (MPS IV) was first described by Dr. Luis Morquio in 1929 in Uruguay. It was also described the same year by Dr. Brailsford in England. While this type is usually known as Morquio Syndrome, it is sometimes known as Morquio-Brailsford Syndrome.

There are two sub-types of mucopolysaccharidosis IV (MPS IV). They are Morquio A and Morquio B. Each sub-type is the result of the deficiency of a certain and unique enzyme. However, there is very little difference in the manifestation of the two sub-types so they will be treated as one here.

## Characteristics of MPS IV

- Growth deficiency - height rarely exceeds 39 inches
- Severe kyphosis (curvature of the spine)
- Flaring of the rib cage
- Frequent upper respiratory infections
- Coarse facial features - broad mouth, widely spaced teeth and upturned nose
- Very short neck
- Legs and arms appear disproportionately long
- Knock knees
- Normal intelligence
- Cloudy corneas after age 10 years
- Progressive deafness during adolescence

Individuals affected by Morquio Syndrome (MPS IV) often have fatal respiratory or cardiac complications during adolescence. There have been individuals with Morquio Syndrome (MPS IV) who have survived into adulthood.

For more information on mucopolysaccharidosis (MPS), please see page 34 of this handbook.

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# MUCOPOLYSACCHARIDOSIS VI

**Synonyms** – MPS VI; Maroteaux-Lamy Syndrome

**When Detectable** – In early childhood or prenatally through enzyme assay of the chorionic villi or amniotic fluid

**Prevalence** – This is a rare syndrome

**Ratio** – 1 male to 1 female

## Description

First described in 1963 by French physicians Dr. Pierre Maroteaux and Dr. Maurice Lamy, this type of mucopolysaccharidosis takes its name from them.

There are three sub-types of mucopolysaccharidosis VI. They are Maroteaux-Lamy - Classic Severe, Maroteaux-Lamy - Intermediate, and Maroteaux-Lamy - Mild. As these names imply, the difference in each sub-type is the severity of the manifestations of the syndrome. Each sub-type shares the following characteristics with differences in severity.

### Characteristics of MPS VI

- Normal development to about age 6 years
- Chest deformity with prominent sternum
- Small stature
- Coarse facial features - thick eyebrows and scalp hair, flat nasal bridge, full cheeks and lips, and large head
- Normal intelligence
- In the severe form - hearing and vision loss and cardiac defects

Individuals with Maroteaux-Lamy - Classic Severe may survive into their late teens or early twenties when they succumb to cardiac or respiratory difficulties. However, individuals with Maroteaux-Lamy-Mild usually have a normal lifespan.

For more information on mucopolysaccharidosis (MPS), please see page 34 of this handbook.

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# MUCOPOLYSACCHARIDOSIS VII

**Synonyms** – MPS VII; Sly Syndrome

**When Detectable** – At birth, in early childhood or prenatally through enzyme assay of the chorionic villi or amniotic fluid

**Prevalence** – Estimated at 1 in 250,000 births

**Ratio** – 1 male to 1 female

## Description

First described in 1973 by American Dr. William Sly for whom the syndrome is named. Mucopolysaccharidosis VII (MPS VII) is one of the least common types of mucopolysaccharidosis.

## Characteristics of MPS VII

- Growth deficiency
- Joint contractures (limitation of the range of motion in the joint)
- Spinal malformations
- Coarse facial features - thick lips, flat nose, heavy eyebrows
- Mental retardation
- Cloudy corneas
- Dislocated hips
- Frequent respiratory illnesses

Due to the fact that mucopolysaccharidosis VII (MPS VII) is one of the most rare and most recently described types of mucopolysaccharidosis, there is little known about the long term prognosis for individuals diagnosed with this type of mucopolysaccharidosis.

For more information on mucopolysaccharidosis (MPS), please see page 34 of this handbook.

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# NEUROFIBROMATOSIS

**Synonyms** – NF; von Recklinghausen Disease; von Recklinghausen’s Neurofibromatosis; Others depending on type.

**When Detectable** – At birth or by age 5 years

**Prevalence** – Varies depending on type

**Ratio** – 1 male to 1 female

## Description

First described in 1882 by German physician Friederich von Recklinghausen.

**Neurofibromatosis 1** (NF1; Peripheral Neurofibromatosis) Estimated to occur in 1 in 4,000 births.

Characteristics of Neurofibromatosis 1

- Multiple café-au-lait spots (coffee-with-milk colored patches on the skin)

- Neurofibromas (benign tumors that grow on the nerves)

- Freckles on the armpits or groin

- Lisch nodules (pigmented specks or clumps on the iris or colored portion of the eye)

- Scoliosis (curvature of the spine) may be present

- Bowing of the legs may be present

- Tumors may develop on the cranial nerves in the brain or on the spinal cord

- Learning disabilities are present in 50% of individuals with NF1

**Neurofibromatosis 2** (NF2; Bilateral Acoustic Neurofibromatosis)

Estimated to occur in 1 in 40,000 births.

Characteristics of Neurofibromatosis 2

- Multiple tumors on the cranial and spinal nerves

- Lesions on the brain and spinal cord

- Hearing loss in the beginning in the individual’s teens or twenties caused by tumors on the auditory nerves

Each child of a parent with neurofibromatosis has a 50% chance of having neurofibromatosis.

Researchers have determined that NF1 and NF2 are caused by different genes located on different chromosomes and are therefore distinct disorders.

It was once widely believed that John Merrick, the so-called “Elephant Man,” had neurofibromatosis. However, Merrick is now believed to have had Proteus Syndrome and not neurofibromatosis.

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# NEURONAL CEROID LIPOFUSCINOSES

**Synonyms** – NCL; Batten Disease; others depending on type

**When Detectable** – Varies depending on type

**Prevalence** – Varies depending on type - over all types estimated at 2 in 100,00 to 4 in 100,000

**Ratio** – 1 male to 1 female

## Description

First described in 1862 by Dr. Christian Stengel in Norway. While there are various names for the different types of neuronal ceroid lipofuscinoses (NCL), Batten Disease which once referred exclusively to the juvenile form is now often used to refer to all the types of NCL.

All forms of NCL result from a buildup of lipopigments in the various tissues of the body. The lipopigments are made up of fat and proteins. The lipopigments form deposits in the brain, eye, muscle, skin and other tissues.

**Infantile NCL** (Santavuori-Haltia disease) - Onset is between the ages of 6 months and 2 years with rapid progression. Characteristics include: failure to thrive, microcephaly, myoclonic jerks (short, shock-like muscle contractions), delayed development with progressive deterioration, and seizures. Infantile NCL is fatal, usually before the fifth birthday.

**Late Infantile NCL** (Jansky-Bielschowsky disease) - Onset is between the ages of 2 and 4 years with rapid progression. Characteristics include: progressive vision loss, loss of muscle coordination, deterioration of mental function and seizures. Late Infantile NCL is fatal, generally between the ages of 8 and 12 years.

**Juvenile NCL** (Batten Disease) - Onset is between the ages of 5 and 8 years with less rapid progression. Characteristics include: progressive vision loss, loss of muscle coordination resulting in clumsiness and seizures. Juvenile NCL is fatal, individuals with Juvenile NCL usually live into their late teens or twenties.

**Adult NCL** (Kufs Disease or Parry's Disease) - Onset is usually after the age of 40 years, with slow progression. Adult NCL does not produce blindness and does not appear to shorten the life expectancy.

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# NOONAN SYNDROME

**Synonyms** – NS; Turner-like Syndrome; Turner Phenotype

**When Detectable** – At birth

**Prevalence** – Estimated at 1 in 1,000 to 1 in 2,500 live births

**Ratio** – 1 male to 1 female

## Description

First described in 1963 by American pediatrician Jacqueline Noonan for whom the syndrome is named.

### Characteristics of Noonan Syndrome

Short stature

Hair extends low onto the neck

Short or webbed neck

Hypertelorism (wide-set eyes)

Eyes slant downward

Mental retardation is present in about 50% of individuals with Noonan Syndrome

Learning disabilities

Epicanthal folds (vertical folds of skin which hide where the upper and lower eyelids meet on either side of the nose)

Heart defects are present in two-thirds of individuals with Noonan Syndrome (most often in the form of pulmonic valve anomalies)

Pectus carinatum (pigeon breast or protruding breast bone) or pectus excavatum (funnel chest or indented breast bone)

Possible cryptorchism (failure of testes to descend) in males

Noonan Syndrome is sometimes referred to as Turner-like Syndrome because individuals with Noonan Syndrome exhibit similar physical characteristics to individuals with Turner Syndrome. Noonan Syndrome affects both males and females while Turner Syndrome affects only females. Despite the similarity of physical characteristics between the two syndromes, Noonan Syndrome and Turner Syndrome are distinct from each other.

Between 30% and 75% of all individuals inherit Noonan Syndrome from a parent. In a majority of cases, the mother is also affected by Noonan Syndrome.

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## PERVASIVE DEVELOPMENTAL DISORDER - NOT OTHERWISE SPECIFIED

**Synonyms** – PDD-NOS; Atypical Autism

**When Detectable** – After age 3 years

**Prevalence** – Estimates range from 2 in 10,000 to 16 in 10,000 but may be equal to that for autism

### Description

Pervasive Developmental Disorder - Not Otherwise Specified (PPD-NOS) is a diagnostic term used when an individual does not meet the diagnostic criteria for one of the other Pervasive Developmental Disorders (see page 9 of this handbook). Individuals with PDD-NOS have social and developmental deficits. PDD-NOS is often called “atypical autism” because individuals diagnosed with PDD-NOS exhibit many of the same characteristics as autism with atypical symptomology and/or subthreshold symptomology Characteristics (while the characteristics of PDD-NOS are the same as those for autism - there may be a difference in severity)

- Difficulty relating to other people

- Limited or abnormal eye contact

- Delayed communication with immature speech rhythms and inappropriate work usage

- May use gestures instead of language to communicate

- Language comprehension is impaired

- Highly sensitive to sensory input - may react indifferently or with emotional outbursts

- Difficulty dealing with changes in environment or schedule

- Inappropriate laughing or giggling may be present

- Stereotypical movements - rhythmic movements may include hand flapping, rocking, spinning, toe walking or finger posturing

- May develop an inappropriate attachment to an object

- Echolalia (repeating phrases said by others)

- Lack of spontaneous or imaginative play

- Unusual interests which may appear obsessive or compulsive

It is important for professionals to eliminate other conditions which may be similar to PDD-NOS before diagnosing PDD-NOS.

For more information on autism spectrum disorders, see page 9 of this handbook.

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# PHENYLKETONURIA

**Synonyms** – PKU; Folling Syndrome; Hyperphenylalanemia; Phenylalanine Hydroxylase Deficiency

**When Detectable** – At birth through blood testing

**Prevalence** – Estimated at 1 in 12,000 to 1 in 15,000 births

**Ratio** – 1 male to 1 female

## Description

First described by Norwegian physician Asborn Folling. The metabolic defect responsible for phenylketonuria was first identified by George A Jervis in 1947. In 1954, German physician H. Bickel first described the treatment of phenylketonuria. American pediatrician Robert Guthrie developed the newborn screening test for phenylketonuria in 1959.

Phenylketonuria is a metabolic disease in which the enzyme phenylalanine hydroxylase is absent or nonfunctional. Without this enzyme the body cannot convert the amino acid phenylalanine to tyrosine. As a result there is an increase of phenylalanine in the blood. It is this increase that is measured in the newborn screening test.

It is very important that infants with phenylketonuria begin a diet low in phenylalanine as soon as possible. Early intervention can halt the harmful effects of excess phenylalanine in the blood. For this reason, most developed countries now screen all newborns for phenylketonuria.

Opinions vary regarding the length of time individuals with phenylketonuria need to remain on a diet low in phenylalanine ranging from 6-10 years to the entire lifespan. However, it is important for women with phenylketonuria to be on the diet low in phenylalanine during pregnancy in order to prevent complications.

Women with untreated phenylketonuria (maternal phenylketonuria) are at high risk for having children with mental retardation, microcephaly, and congenital heart defects but not phenylketonuria.

## Characteristics of Untreated Phenylketonuria

- Severe vomiting and seizures during the first weeks of life
- A distinctive smell, often described as “mousy”
- Extremely dry skin
- Microcephaly (small head size)
- Behavioral difficulties
- Mental retardation

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# PRADER-WILLI SYNDROME

**Synonyms** – PWS; Prader-Labhart-Willi Syndrome; Willi-Prader Syndrome; Hypotonia-Hypogonadism-Hypomentia-Obesity Syndrome (HHHO Syndrome)

**When Detectable** – Shortly after birth

**Prevalence** – Estimated at 1 in 12,000 to 1 in 15,000

**Ratio** – 1 male to 1 female

## Description

Described in 1956 by Swiss pediatricians Andrea Prader and H. Willi for whom the syndrome is named. However, the syndrome may have actually been first described in 1887 by English physician J. Langdon Down, who first described Down Syndrome. Down referred to the syndrome as “polysarcia.”

## Characteristics of Prader-Willi Syndrome

Hypotonia (low muscle tone, sometimes described as “floopy” in appearance) at birth

Poor sucking and swallowing reflexes in the first months of life

Cry may be weak

Failure to thrive in infancy

Prominent forehead

Poorly formed ears

Mouth with a thin upper lip and down-turned corners

Developmental delays

Weight gain that is rapid or excessive between the ages of 1 and 6 years

Hyperphagia (compulsive overeating)

Obsession with food may include “food-seeking” behavior

Mental retardation - ranging from mild to moderate

Delayed or diminished puberty

Hypogonadism (small testicles), possible undescended testicles, hypoplastic (short to absent) scrotum, small penis in males

Poorly formed labia and menses that are scant or absent in females

Behavioral difficulties - may include obsessive or compulsive behaviors

Obesity

The restriction of access to food and monitoring of food intake is vitally important for individuals with Prader-Willi Syndrome. Individuals with Prader-Willi Syndrome have an excessive appetite but have a decreased need for caloric intake. Morbid obesity is a major medical concern for individuals with Prader-Willi Syndrome.

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Prader-Willi Alliance for Research  
28 Vesey Street  
Suite 2104  
New York, NY 10007  
Phone (212) 332-0970  
www.p-war.org

Prader-Willi Syndrome Association  
(USA)  
5700 Midnight Pass Road  
Sarasota, FL 34242  
Phone (941) 312-0400 or  
1-800-926-4797  
www.pwsausa.org

# RETT SYNDROME

**Synonyms** – RS; Rett’s Syndrome; Rett’s Disorder

**When Detectable** – By age 48 months; may also be detected with chromosomal testing

**Prevalence** – Estimated at in 1 in 10,000 to 1 in 15,000 births making it rare and much less common than autism.

**Ratio** – Specific to females

## Description

First described in 1966 by Dr. Andreas Rett in Austria, the syndrome bears his name. However, Rett Syndrome was not widely known in the medical community until 1983.

Individuals with Rett Syndrome develop normally until sometime between the ages of 6 and 18 months. The following characteristics apply to post-onset individuals.

## Characteristics of Rett Syndrome

- Normal head circumference at birth
- Head growth slows as compared to body growth between the ages of 5 months and 4 years
- Loss of purposeful hand movements which are replaced by repetitive hand movements such as “hand washing” movements, wringing and clapping which may become almost constant during waking hours
- Bruxism (teeth grinding) and difficulty chewing
- Language skills do not develop
- Shaking of the limbs and torso
- Unsteady, stiff-legged gait
- Breathing difficulties may include hyperventilation, apnea, air swallowing and breath holding
- Seizures
- Scoliosis (curvature of the spine)
- Mental retardation usually in the severe to profound range
- Regression of cognitive skills
- Regression of social skills
- Regression of motor skills
- Regression of behavioral skills

For more information on autism spectrum disorders, see page 9 of this handbook.

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Autism Society of America  
7910 Woodmont Avenue  
Suite 300  
Bethesda, MD 20814-3067  
Phone (301) 657-0881 or  
1-800-3-AUTISM or  
1-800-328-8476  
www.autism-society.org

International Rett Syndrome  
Association (IRSA)  
9121 Piscataway Road  
Clinton, MD 20735  
Phone (301) 856-3334 or  
1-800-818-RETT or  
1-800-818-7388  
www.rettsyndrome.org

# SPINA BIFIDA

**Synonyms** – SB; Neural Tube Defect

**When Detectable** – At birth or prenatally through ultrasound or amniocentesis

**Prevalence** – Estimated at 1 in 2,000 live births

**Ratio** – 1 male to 1 female

## Description

Spina bifida is a form of neural tube defect (the other forms are anencephaly and encephalocele) in which there is abnormal development of the spinal cord. Spina bifida means “split spine.” There are three types of spina bifida each with a unique set of characteristics.

## Spina Bifida Cystica Myelomeningocele

The most serious form of spina bifida  
Spinal cord along with spinal fluid and spinal nerves protrudes from an opening in the spine and are enclosed by a sac or cyst  
Surgery is generally performed in the first 24 to 48 hours of life  
Nerve damage may lead to paralysis  
Difficulties with bowel and bladder control  
Hydrocephalus

## Spina Bifida Cystica Meningocele

Spinal fluid protrudes from an opening in the spine and is enclosed by a sac or cyst  
No spinal nerves or part of the spinal cord protrude from the spine  
Possible nerve damage may lead to paralysis and difficulties with bowel or bladder control

## Spina Bifida Occulta

The mildest form of spina bifida  
Occulta means “hidden”  
One or more vertebrae is malformed and covered by a layer of skin  
Often there are no symptoms and the condition is revealed by back x-rays  
A sensitive dimple along the spine which may have long dark hairs might appear

While no one is certain what causes spina bifida, women who take a folic acid supplement before and during pregnancy are less likely to have a child with spina bifida. Since spina bifida occurs in the 25th day of pregnancy it is important for women to begin a folic acid regimen before becoming pregnant.

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Spina Bifida Association of America  
4590 MacArthur Boulevard, NW  
Suite 250  
Washington, D.C. 20007-4226  
Phone (202) 944-3285 or  
1-800-621-3141  
www.sbaa.org

March of Dimes  
P.O. Box 1657  
Wilkes-Barre, PA 18703-1657  
Phone 1-800-MODIMES  
1-800-367-6630  
www.modimes.org

Spina Bifida and Hydrocephalus  
Association of Canada  
#977-167 Lombard Avenue  
Winnipeg, Manitoba,  
Canada R3B 0V3  
Phone (204) 925-3650  
www.sbhac.ca

# SPINAL MUSCULAR ATROPHY

**Synonyms** – SMA; Others depending on type

**When Detectable** – Varies according to type

**Prevalence** – Varies according to type - Overall forms estimated at 1 in 6,000 to 1 in 10,000 live births

**Ratio** – 1 male to 1 female

## Description

Spinal Muscular Atrophy affects the nerve cells which control voluntary muscle movement. Involuntary muscles are not affected, intelligence is normal or above and hearing and vision are not affected. There are four types of spinal muscular atrophy.

**Spinal Muscular Atrophy Type 1** (SMA Type 1; Spinal Muscular Atrophy Type 1 - Acute or Severe, Werdnig Hoffman Disease) - SMA Type 1 is usually diagnosed in the first six months of life. Individuals with SMA Type 1 are usually never able to lift their heads or sit without support. Difficulties with breathing, sucking and swallowing are present. SMA Type 1 is usually fatal before age 2 years and often before age 1 year.

**Spinal Muscular Atrophy Type 2** (SMA Type 2; Spinal Muscular Atrophy Type 2 - Chronic) - SMA Type 2 is usually diagnosed before the age of 2 years with a majority of cases diagnosed before 15 months of age. Individuals with SMA Type 2 generally learn to sit unaided but may be unable to assume a sitting position without assistance and require assistance to stand or walk. Feeding and swallowing difficulties may or may not be present. Some individuals with SMA Type 2 may survive past childhood.

**Spinal Muscular Atrophy Type 3** (SMA Type 3; Spinal Muscular Atrophy Type 3 - Mild, Kugelberg-Welander Disease) - SMA Type 3 is usually diagnosed between the ages of 5 to 15 years of age. This is the mildest of the childhood onset Spinal Muscular Atrophies. Individuals with SMA Type 3 are able to sit and stand unaided, although progressive muscle weakness may require wheelchair use. Individuals with SMA Type 3 may survive into adulthood with fatalities most often occurring from respiratory complications.

**Spinal Muscular Atrophy Type 4** (SMA Type 4; Spinal Muscular Atrophy Type 4 - Adult Onset) - SMA Type 4 onset begins around age 35. Symptoms usually begin in the hands, feet or tongue and progression is slow.

It is estimated that 1 in 40 to 1 in 80 people is a carrier of the Spinal Muscular Atrophy gene. Each child conceived by two carriers has a 1 in 4 chance of developing Spinal Muscular Atrophy.

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Families of Spinal Muscular  
Atrophy  
P.O. Box 196  
Liberty, IL 60048-0196  
Phone (847) 367-7620 or  
1-800-886-1762  
[www.fsma.org](http://www.fsma.org)

Muscular Dystrophy Association  
(MDA) - USA  
National Headquarters  
3300 East Sunrise Drive  
Tucson, AZ 85718  
Phone 1-800-572-1717  
[www.mdaua.org](http://www.mdaua.org)

# TOURETTE SYNDROME

**Synonyms** – TS; Tourette’s Syndrome; Gilles de la Tourette Syndrome; GTS; Tourette’s Disorder; Chronic Multiple Tics; Chronic Motor Tics

**When Detectable** – Onset is usually between the ages of 2 and 15 years and typically before age 21 years

**Prevalence** – Estimated at 1 in 2,000 live births

**Ratio** – Estimates range from 3 males to 1 female to 4 males to 1 female

## Description

First described in 1885 by French neurologist Georges Gilles de la Tourette for whom the syndrome is named.

### Characteristics of Tourette Syndrome

Multiple motor tics which may include:

Simple Tics - eye blinking, head jerking, shoulder shrugging or facial grimacing

Complex Tics - jumping, smelling, twirling, touching things or other people, compulsive behaviors or very rarely self-injurious behaviors

One or more vocal tics which may include:

Simple Tics - throat clearing, yelping, sniffing, coughing, tongue clicking or other noises

Complex Tics - using words or phrases out of context, echolalia (repeating a sound, word or phrase just heard), palilalia (repeating one’s own words), or rarely coprolalia (speaking socially unacceptable or obscene words)

Tics occurring many times daily nearly every day

Changes in the number, frequency and type of tics

Possible obsessive, compulsive or ritualistic behaviors

Possible Attention Deficit Hyperactivity Disorder

Possible learning disabilities

Possible sleep disorders

Possible difficulties with impulse control

It is possible for individuals with Tourette Syndrome to experience periods of remission in which the tics decrease in severity or frequency. The symptoms of Tourette Syndrome tend to be worse during puberty and to stabilize during adulthood.

An estimated 15% of all children exhibit a transient tic during childhood, these tics are not indicative of Tourette Syndrome.

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Tourette Syndrome Association,  
Inc.  
42-40 Bell Boulevard  
Bayside, NY 11361-2820  
Phone (718) 224-2999  
www.tsa-usa.org

WE MOVE (Worldwide Education  
and Awareness of Movement  
Disorders)  
204 West 84th Street  
New York, NY 10024  
Phone 1-800-437-MOV2 or  
1-800-437-6682  
www.wemove.org

Tourette Syndrome Foundation of  
Canada  
#206, 194 Jarvis Street  
Toronto, Ontario M5B 2B7  
Phone (416) 861-8398  
www.tourette.ca

# TRAUMATIC BRAIN INJURY

**Synonyms** – TBI; Brain Trauma

**Prevalence** – Each year an estimated 1.5 million individuals in the United States sustain a traumatic brain injury. Of those, 1 million are treated and released from hospital emergency departments, 230,000 are hospitalized and survive, and 50,000 die.

**Ratio** – Estimated at 2 males to 1 female

## Description

Traumatic brain injury is generally divided into two events - the primary injury (a blow or jolt to the head) and secondary injuries which result from the primary injury.

## Types of Primary Injuries

Skull Fracture - break in the bone of the skull

Contusions (bruises) - often occur under the area of impact but may also occur where the brain is forced against the bony ridges inside the skull

Hematomas (blood clots) - result when small blood vessels are broken and may occur between the skull and the brain (epidural or subdural hematoma) or in the brain itself (intracerebral)

Lacerations - tears caused by the brain moving across the bony ridges inside the skull

Diffuse Axonal Injury - shearing injury to the brain's long connecting nerve fibers or axons caused by the rotation and shifting of the brain

## Types of Secondary Injuries

Hematoma (blood clots)

Increased intracranial pressure

Intracranial infection

Brain swelling

Cerebral vasospasm

Epilepsy

Overall the main causes of traumatic brain injuries are vehicle accidents, firearms, and falls. For children, the main causes of brain injury are falls, abuse (such as "Shaken Baby Syndrome"), recreation accidents and vehicle accidents.

The effect of a traumatic brain injury can range from mild to fatal. It is estimated that there are 5.3 million Americans living with a traumatic brain injury related disability. Prevention of traumatic brain injuries is becoming a major public health issue.

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Brain Injury Association of America  
105 North Alfred Street  
Alexandria, VA 22314  
Phone (702) 236-6000 or  
Family Helpline at  
1-800-444-6443  
www.biausa.org

Brain Trauma Foundation  
523 East 72nd Street  
New York, NY 10021  
Phone (212) 112-0608  
www.braintrauma.org

# TUBEROUS SCLEROSIS

**Synonyms** – TS: Tuberous Sclerosis Complex; TSC; Tuberous Sclerosis Syndrome; Andenoma Sebaceum; Bourneville Disease; Epiloia

**When Detectable** – In infancy

**Prevalence** – Estimated at 1 in 6,000 to 1 in 50,000 live births

**Ratio** – 1 male to 1 female

## Description

First described in 1862 by German physician Friederich von Recklinghausen who was also the first to describe neurofibromatosis.

### Characteristics of Tuberous Sclerosis

Benign (non-cancerous) tumors often on the brain, eyes, heart, kidney, skin and lungs

Seizures

Angiofibroma (reddish bumps in a butterfly pattern across the cheeks)

More than 3 hypomelanotic macules (“white spots” or “ash leaf” spots in which the skin has no pigmentation)

Café-au-lait (coffee-with-milk) spots

Shagreen patches (areas of the skin where collagen collects that appear yellowish-brown in color and are slightly elevated)

Tooth enamel defects

Mental retardation may be present

Learning disabilities may be present

Individuals with tuberous sclerosis fall into 1 of 3 categories.

Non-Affected individuals have none of the debilitating effects of the disease.

Mildly-Affected individuals have structural brain anomalies, may have seizures, may have learning disabilities, may have complications from tumors on internal organs.

Severely-Affected individuals usually have mental retardation ranging from moderate to severe, behavioral difficulties and more pronounced symptoms of the disease.

It is important for individuals who are classified as non-affected to be correctly diagnosed as it is possible for them to pass tuberous sclerosis to their children.

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Tuberous Sclerosis Alliance  
801 Roeder Road  
Suite 750  
Silver Spring, MD 20910  
Phone 1-800-225-6872  
[www.tsalliance.org](http://www.tsalliance.org)

Epilepsy Foundation  
4351 Garden City Drive  
Landover, MD 20785-7223  
Phone 1-800-332-1000  
[www.epilepsyfoundation.org](http://www.epilepsyfoundation.org)



# TURNER SYNDROME

**Synonyms** – Turner’s Syndrome; XO Syndrome

**When Detectable** – At birth through adulthood

**Prevalence** – Estimated at 1 in 2,500 female births

**Ratio** – Specific to females

## Description

Turner Syndrome was first described by American doctor Henry H. Turner in 1938. The syndrome carries Dr. Turner’s name. The chromosomal cause of Turner Syndrome was identified in 1959 by Dr. C.E. Ford.

## Characteristics of Turner Syndrome

- Short stature - average height of 4 feet, 7 inches
- Underdeveloped ovaries
- Possible difficulties with the ears, eyes, heart, kidney or thyroid
- Low set ears
- Low hairline
- Webbed neck
- Broad chest
- Puffiness of the hands and feet
- Infertility

Turner Syndrome does not cause mental retardation. However, verbal learning tends to come more easily to those affected by Turner Syndrome than do mathematical or spatial learning skills. Individuals with Turner Syndrome may also have a learning disability.

Turner Syndrome does not affect the lifespan unless there are serious medical issues related to abnormalities of the heart, kidneys or thyroid.

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Turner Syndrome Society  
14450 T.C. Jester  
Suite 260  
Houston, TX 77014  
Phone (832) 249-9988 or  
1-800-365-9944  
www.turner-syndrome-us.org

Human Growth Foundation, Inc.  
997 Glen Cove Avenue  
Glen Head, NY 11545  
Phone (516) 671-4041 or  
1-800-451-6434  
www.hgfound.org

Turner’s Syndrome Society of  
Canada  
814 Glencairn Avenue  
Toronto, Ontario M6B 2A3  
Phone (416) 781-2086  
www.turnersyndrome.ca

# WILLIAMS SYNDROME

**Synonyms** – WS; Williams-Bueren Syndrome; WBS; Elfin Facies Syndrome; Efin Facies with Hypercalcemia

**When Detectable** – At birth through childhood

**Prevalence** – Estimated at 1 in 20,000 live births

**Ratio** – 1 male to 1 female

## Description

First described in 1961 by New Zealand physician J.C.P. Williams for whom the syndrome is named.

### Characteristics of Williams Syndrome

Characteristic facial features including small upturned nose, long philtrum (ridges between the nose and mouth), wide mouth, full lips, small chin, prominent earlobes, widely spaced teeth and puffiness around the eyes

Low birth-weight

Slow weight gain during infancy

Possible failure to thrive

Possible feeding problems in infancy

Heart or blood vessel problems often narrowing of the arteries

Hypercalcemia (elevated blood calcium levels) which may lead to extreme irritability or colic-like behavior in infancy

Hyperacusis (sensitive hearing)

Personality that is overly friendly or excessively social

Developmental delays

Mental retardation ranging from mild to severe

Individuals with Williams Syndrome generally have strong skills in the areas of auditory rote memory, language and speech. Individuals with Williams Syndrome tend to have weaker skills in areas such as fine motor skills and visuospatial construction.

The characteristic facial features of Williams Syndrome tend to become more pronounced as individuals with the syndrome age.

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The Williams Syndrome  
Association  
P.O. Box 297  
Clawson, MI 48017-0297  
Phone (248) 541-3630  
www.williams-syndrome.org

Williams Syndrome Foundation  
University of California  
Irvine, CA 92679-2310  
Phone (949) 824-7259  
www.wsf.org

# ZELLWEGER SYNDROME

**Synonyms** – Cerebrohepatorenal Syndrome; CHRS

**When Detectable** – At birth

**Prevalence** – Estimated at 1 in 25,000 to 1 in 100,000 births

**Ratio** – 1 male to 1 female

## **Description**

First described in 1964 by Dr. Hans V. Zellweger in America, the syndrome bears his name.

### Characteristics of Zellweger Syndrome

Prenatal growth deficiencies

Low birth weight

Hypotonia (lack of muscle tone) may result in “floppy infant” appearance - some infants are unable to move

Mental retardation

Enlarged liver

Characteristic facial features - high, bulging forehead, round face, wide-set eyes, puffy eye lids, ears poorly formed and small jaw

Inability to suck and/or swallow

Glaucoma, corneal clouding, and cataracts

Heart defects

Cysts on the kidneys

Seizures may occur

Extra care is needed to control respiratory infections. Individuals affected by Zellweger Syndrome usually survive only a few months. Death can be caused by respiratory distress, gastrointestinal bleeding or liver failure.

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United Leukodystrophy  
Foundation (ULF)  
2304 Highland Drive  
Sycamore, IL 60178  
Phone 1-800-728-5483  
www.ulf.org

# OVERVIEW OF ADA, IDEA, AND SECTION 504

Americans with Disabilities Act of 1990 (ADA)

Individuals with Disabilities Education Act (IDEA)

Section 504 of the Rehabilitation Act of 1973

## Type / Purpose

A civil rights law to prohibit discrimination solely on the basis of disability in employment, public services and accommodations.

An education act to provide federal financial assistance to state and local education agencies to guarantee special education and related services to eligible children with disabilities.

A civil rights law to prohibit discrimination on the basis of disability in programs and activities, public and private, that receive federal financial assistance.

## Who is Protected?

Any individual with a disability who: (1) has a physical or mental impairment that substantially limits one or more life activities: or (2) has a record of such an impairment: or (3) is regarded as having such an impairment. Further, the person must be qualified for the program, services or job. Major life activities include walking, seeing, hearing, speaking, breathing, learning, working, caring for oneself, and performing manual tasks.

Children ages 3-21 years who are determined by a multidisciplinary team to be eligible within one or more of 13 specific disability categories and who need special education and related services. Categories include autism, deafness, deaf-blindness, hearing impairments, mental retardation, multiple disabilities, orthopedic impairments, other health impairments, serious emotional disturbance, specific learning disabilities, speech or language impairments, traumatic brain injury, and visual impairments.

Any person who (1) has a physical or mental impairment that substantially limits one or more major life activities, (2) has a record of such an impairment, or (3) is regarded as having such an impairment. Major life activities include walking, seeing, hearing, speaking, breathing, learning, working, caring for oneself, and performing manual tasks.

## Funding to Implement Requirements?

No, but limited tax credits may be available for removing architectural or transportation barriers. Also, many federal agencies provide grant funds to support training and to provide technical assistance to public and private institutions.

Yes. IDEA provides federal funds under Parts B and H to assist states and local education agencies in meeting IDEA requirements to serve infants, toddlers, children, and youth with disabilities.

No. State and local jurisdictions have fiscal responsibility. IDEA funds may not be used to serve children found eligible only under Section 504.

# OVERVIEW OF ADA, IDEA, AND SECTION 504

Americans with Disabilities Act of 1990 (ADA)

Individuals with Disabilities Education Act (IDEA)

Section 504 of the Rehabilitation Act of 1973

## Responsibility to Provide a Free, Appropriate Public Education (FAPE)?

Not directly. However, (1) ADA protections apply to nonsectarian private schools, but not to organizations or entities controlled by religious organizations; (2) ADA provides additional protection in combination with actions brought under IDEA and Section 504. Reasonable accommodations are required for eligible students with a disability to perform essential functions of a job. This applies to any part of the special education program that may be community-based and involve job training/ placement.

Yes. A FAPE is defined to mean special education and related services. Special education means “specifically designed instruction, at no cost to the parents, to meet the unique needs of the child with a disability . . .” Related services are provided if students require them in order to benefit from specially designed instruction. States are required to ensure the provision of “full educational opportunity” to all children with disabilities.

IDEA requires the development of an Individualized Education Program (IEP) document with specific content and a required number of specific participants at an IEP meeting.

Yes. An “appropriate” education means an education comparable to that provided to students without disabilities. This may be defined as regular or special education services. Students can receive related services under Section 504 even if they are not provided any special education.

Section 504 does require the development of a plan, although a written document is not mandated. The Individualized Education Plan (IEP) mandated by IDEA may be used for the Section 504 written plan. Many experts recommend that a group of persons knowledgeable about the students convene and specify the agreed-upon services.

## Procedural Safeguards?

The ADA does not specify procedural safeguards related to special education. However, it does detail the administrative requirements, complaint procedures, and the consequences for noncompliance, related to both services and employment.

IDEA requires written notice to parents regarding identification, evaluation and/or placement. Further, written notice must be made prior to any change in placement. The Act delineates the required components of the written notices.

Section 504 requires notice to parents regarding identification, evaluation, and/or placement. A written notice is recommended. Notice must be made only before a “significant change” in placement. Following IDEA procedural safeguards is one way to meet Section 504 mandates.

# OVERVIEW OF ADA, IDEA, AND SECTION 504

Americans with Disabilities Act of 1990 (ADA)

Individuals with Disabilities Education Act (IDEA)

Section 504 of the Rehabilitation Act of 1973

## Evaluation / Placement Procedures?

The ADA does not specify evaluation and placement procedures. However, it does specify provision of reasonable accommodations for eligible students across educational activities and settings. Reasonable accommodations may include, but are not limited to, redesigning equipment, assigning aides, providing written communication in alternative formats, altering existing facilities, and building new facilities.

A comprehensive evaluation is required. A multidisciplinary team evaluates the child, and parental consent is required before an initial evaluation. IDEA requires that reevaluation be conducted at least every three years. A reevaluation is not required before a significant change in placement.

For evaluation and placement decisions, IDEA requires that more than a single procedure or information source be used; that information from all sources be documented and carefully considered; that the eligibility decision be made by a group of persons who know about the student, the evaluation date, and placement options; and that the placement decision serves the student in the least restrictive environment. An IEP review meeting is required before any change in placement.

Unlike IDEA, Section 504 requires only notice, not consent, for evaluation. It is recommended that districts obtain parental consent.

Like IDEA, evaluation and placement procedures under Section 504 require that information be obtained from a variety of sources in the area of concern; that all data are documented and considered; and that decisions are made by a group of persons knowledgeable about the student, evaluation data, and placement options. Section 504 requires periodic reevaluations, but does not specify any timelines for reevaluations. Section 504 requires that students be educated with their nondisabled peers to the maximum extent appropriate. Section 504 does not require a meeting for any change in placement.

## Due Process?

People with disabilities have the same remedies that are available under Title VII of the Civil Rights Act of 1964, as amended in 1991. Thus, individuals who are discriminated against may file a complaint with the relevant federal agency or sue in federal court. Enforcement agencies encourage informal mediation and voluntary compliance.

IDEA delineates specific requirements for local education agencies to provide impartial hearings for parents who disagree with the identification, evaluation or placement of a child.

Section 504 requires local education agencies to provide impartial hearings for parents who disagree with the identification, evaluation, or placement of a student. It requires that parents have an opportunity to participate in the hearing process and to be represented by counsel. It is recommended that districts develop policy and procedures.

# SELECTED RESOURCES

The following list of selected resources related to developmental disabilities along with many other resources are available through the Wegner Health Science Information Center. To access any of these resources visit the Wegner Health Science Information Center at 1400 West 22nd Street, Sioux Falls, South Dakota, 57103 or call the Center for Disabilities at (605) 357-1439 or toll-free 1-800-658-3080 (Voice/TTY). You can also visit your local library for help in locating resources.

Adaptation to Chronic Childhood Illness by Robert J. Thompson, Jr. and Kathryn E. Gustafson, 1996

Adolescents with Down Syndrome: Toward a More Fulfilling Life edited by Siegfried M. Pueschel and Maria Sustrova, 1997

Answering Your Questions About Spina Bifida by The Spina Bifida Program, Children's National Medical Center, 1995

"Ask Me About Asperger's Syndrome" videotape produced by Michael Thompson Productions, 2000

Asperger Syndrome: A Guide for Educators and Parents by Brenda Smith Myles and Richard L. Simpson, 1998

Asperger Syndrome and Difficult Moments: Practical Solutions for Tantrums, Rage and Meltdown by Brenda Smith Myles and Jack Southwick, 1999

Asperger Syndrome and Sensory Issues: Practical Solutions for Making Sense of the World by Brenda Smith Myles, et al., 2000

Asperger Syndrome in the Family: Redefining Normal by Liane Holiday Willey, 2001

An Atlas of Epilepsy by D.F. Smith, et al., 1998

Attention Deficit Disorder by Alvin Silverstein, Virginia Silverstein and Laura Silverstein Nunn, 2001

"Attention Deficit Hyperactivity Disorder" videotape produced by Interactive Teaching Network, University of Georgia, 1997

Autism & PDD: Picture Stories & Language Activities by Patricia Snair Koski, 1998

Autism & PDD: Social Skills Lessons by Pam Britton Reese and Nena C. Challenner, 1999

Babies with Down Syndrome: A New Parents' Guide edited by Karen Stray-Gundersen, 1995

Be Good to Eddie Lee by Virginia Fleming and illustrated by Floyd Cooper, 1993 (Down Syndrome)

Beyond the Wall: Personal Experiences with Autism and Asperger Syndrome by Stephen Shore, 2001

Blue Bottle Mystery: An Asperger Adventure by Kathy Hoopmann, 2001

# SELECTED RESOURCES

Breaking Autism's Barriers: A Father's Story by Bill Davis as told to Wendy Goldband Schunick, 2001

"Breakthroughs: How to Reach Students with Autism" videotape produced by Jeff Schultz, 1998

Buster and the Amazing Daisy by Nancy Ogaz with illustrations by Patricia Shubeck, 2002 (Autism)

Caring for Children with Cerebral Palsy: A Team Approach edited by John P. Dormans and Louis Pelegrino, 1998

Challenging Behavior and Autism: Making Sense - Making Progress: A Guide to Preventing and Managing Challenging Behaviour for Parents and Teachers by Philip Whitaker, 2001

Charcot-Marie-Tooth Disorders: A Handbook for Primary Care Physicians edited by Gareth J. Parry, 1995

Charlsie's Chuckle by Clara Widess Berkus, 1992 (Down Syndrome)

Children with Autism: A Parent's Guide edited by Michael D. Powers, 2000

Children with Cerebral Palsy: A Parents' Guide edited by Elaine Geralis, 1991

Children with Disabilities by Mark L. Batshaw, 1997

Children with Tourette Syndrome: A Parents' Guide edited by Tracy Haerle, 1992

Children with Spina Bifida: A Parent's Guide edited by Marlene Lutkenhoff, 1999

Cognitive Coping, Families, and Disability edited by Ann P. Turnbull, et al., 1993

Communication Strategies for People with Developmental Disabilities: Issues from Theory and Practice edited by Ken Linfoot, , 1994

Confronting Traumatic Brain Injury: Devastation, Hope and Healing by William J. Winslade, 1998

Coping with Mild Traumatic Brain Injury by Diane Roberts Stoler and Barbara Albers Hill, 1998

Count Us In: Growing Up with Down Syndrome by Jason Kingsley and Mitchell Levitz, 1994

Creating Collaborative IEPs: A Handbook edited by Kate Wallace McCoy, 1998

Cystic Fibrosis edited by Margaret E. Hodson and Duncan M. Geddes, 2000

Cystic Fibrosis: Medical Care by David M. Orenstein, Beryl J. Rosenstein and Robert C. Stern, 2000



# SELECTED RESOURCES

- Dear Charlie: A Grandfather's Love Letter: A Guide to Your Life with Autism by Earl P. Martin, Jr., 1999
- Developing Leisure Time Skills for Persons with Autism: A Practical Approach for Home, School and Community by Phyllis Coyne, Colleen Nyberg and Mary Lou Vandenburg, 1999
- Developmental Disabilities in Infancy and Childhood edited by Arnold J. Capute and Pasquale J. Accardo, 1996
- Dictionary of Developmental Disabilities Terminology by Pasquale J. Accardo, Barbara Y. Whitman, et al., 1996
- "A Different Way of Learning: The Employee with a Learning Disability" videotape produced by Learning Disabilities Association of Ontario, 1997
- Down Syndrome Birth to Adulthood: Giving Families an Edge by John E. Rynders and J. Margaret Horrobin, 1996
- Eating an Artichoke: A Mother's Perspective on Asperger Syndrome by Echo R. Fling, 2000
- Educating Children with Multiple Disabilities: A Transdisciplinary Approach by Fred P. Orelove and Dick Sobsey, 1996
- "Educating Peter" videotape produced by Ambrose Video Publishing, 1993 (Down Syndrome)
- "Effective Education Adapting to Include All Students," "Essential Characteristics of Inclusive Schools," and "Valuing Parents in the IEP Process Making Decisions About What and Where Students Learn" set of three videotapes produced by the South Dakota Department of Education and Cultural Affairs, 2000
- "Ennis' Gift: A Film about Learning Differences" videotape produced by Hello Friend/Ennis William Cosby Foundation, 2000
- Epilepsy and Developmental Disabilities edited by Orrin Devinsky and Lauren E. Westbrook, 2002
- Everybody is Different: A Book for Young People who have Brothers or Sisters with Autism by Fiona Bleach, 2001
- "Faces Yet to Come" videotape produced by American Indian Institute, University of Oklahoma, 1997 (Fetal Alcohol Syndrome)
- Facing Autism: Giving Parents Reasons for Hope and Guidance for Help by Lynn M. Hamilton, 2000
- Families, Disability and Empowerment: Active Coping Skills and Strategies for Family Interventions edited by George H.S. Singer and Laurie E Powers, 1993
- Family Articles About Traumatic Brain Injury by Angela Tipton Dikengil, 1994

# SELECTED RESOURCES

“Family Perspectives on Inclusion Across the Lifespan” videotape produced by the Center for Aging Persons with Developmental Disabilities, Institute for the Study of Developmental Disabilities, Indiana University, 1993

Fetal Alcohol Syndrome: Diagnosis, Epidemiology, Prevention, and Treatment Institute of Medicine, 1996

Fetal Alcohol Syndrome: A Guide for Families and Communities by Ann Streissguth, 1997

Fragile Success: Ten Autistic Children, Childhood to Adulthood by Virginia Walker Sperry, 2001

The Fragile X Child edited by Betty B. Schopmeyer and Fonda Lowe, 1992

“The Fragile X Mystery” videotape produced by Ambrose Video Publishing, 1990

A Guide to Cystic Fibrosis for Parents and Children by James C. Cunningham and Lynn M. Taussig, 1988

Handbook of Autism and Pervasive Developmental Disorders edited by Donald J. Cohen and Fred R. Volkmar, 1997

Handbook of Developmental Disabilities: Resources for Interdisciplinary Care edited by Lisa A. Kurtz, et al., 1996

Handbook of Epilepsy by Thomas R. Browne and Gregory L. Holmes, 2000

Help is on the Way: A Child’s Book about ADD by Marc A. Nemiroff, Jane Annunziata and illustrated by Margaret Scott, 1998

Helping Your ADD Child: Hundreds of Practical Solutions for Parents and Teachers of ADD Children and Teens (with or without hyperactivity) by John F. Taylor, 2001

A History of Childhood and Disability by Philip L. Sanford, 1996

I Am Special: Introducing Children and Young People to Their Autistic Spectrum Disorder by Peter Vermeulen, 2000

Ian’s Walk: A Story about Autism by Laurie Lears, illustrated by Karen Ritz, 1998

“The IEP: A Tool for Realizing Possibilities” videotape produced by Peak Parent Center, 1998

“I’m the Big Sister Now” by Michelle Emmert and illustrated by Gail Owens, 1989 (Cerebral Palsy)

Implementing the Americans with Disabilities Act: Rights and Responsibilities of All Americans edited by Lawrence O. Gostin and Henry A. Beyer, 1993

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Innovations in Family Support for People with Learning Disabilities edited by Peter Mittler and Helle Mittler, 1994

Inside Out: What Makes a Person with Social Cognitive Deficits Tick? by Michelle Garcia Winner, 2000

“Just Being Kids: Supports and Services for Infants and Toddlers and Their Families in Everyday Routines, Activities, and Places” videotape produced by Colorado Department of Education, 2001

“Just Friends” videotape produced by James Brodie Productions, Inc., 1997

Just This Side of Normal: Glimpses into Life with Autism by Elizabeth King Gerlach, 1999

Keys to Parenting a Child with Down Syndrome by Marlene Targ Brill, 1993

Kids Explore the Gifts of Children with Special Needs by Westridge Young Writers Workshop, 1994

“Last Call: The Sobering Truth About FAS,FAE” videotape produced by Tom Monson and Advantage Source, 2001

Learning Disabilities: Lifelong Issues edited by Shirley C. Cramer and William Ellis, 1996

Learning to Slow Down and Pay Attention by Kathleen G. Nadeau, Ellen B. Dixon and illustrated by John R. Rose, 1997

“Look What You’ve Done!: Stories of Hope and Resilience” videotape produced by Noel Gunther, 1997 (Learning Disabilities)

Making Sense of Art: Sensory-Based Art Activities for Children with Autism, Asperger Syndrome and Other Pervasive Developmental Disorders by Sandra R. Davalos, 1999

Management of Genetic Syndromes edited by Suzanne B. Cassidy and Judith E. Allanson, 2001

Meal Preparation: Teaching Individuals with Developmental Disabilities to Cook by Linda R. Harper, 1998

Medical Facts About Spina Bifida by Kennedy Krieger Institute, 1995

Mental Retardation: Nature, Cause, and Management by George S. Baroff with J. Gregory Olley, 1999

A Mind of its Own: Tourette’s Syndrome: A Story and Guide by Ruth Dowling Bruun and Bertel Bruun, 1994

The Misunderstood Child: Understanding and Coping with Your Child’s Learning Disabilities by Larry B. Silver, 1998

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Muscular Dystrophy in Children: A Guide for Families by Irwin M. Siegel, 1999

Muscular Dystrophy: The Facts by Alan E.H. Emergy, 1994

My Buddy by Audrey Osofsky and illustrated by Ted Rand, 1994 (Muscular Dystrophy)

My Social Stories Book by Carol Gray, 2001 (Autism)

“Normal for Us: The Miller Twins” videotape produced by Oregon Public Broadcasting, 2002 (Spinal Muscular Atrophy)

The Oasis Guide to Asperger Syndrome: Advice, Support, Insights, and Inspiration by Patricia Romanowski Bashe and Barbara L. Kirby, 2001

On the Outside: Extraordinary People in Search of Ordinary Lives edited by Julie Pratt, 1998

“Oral Hygiene for Persons with Developmental Disabilities” videotape produced by University of Washington, School of Dentistry, 1996

Parent Articles about ADHD edited by Clare B. Jones, et al., 1999

A Parent’s Guide to Down Syndrome: Toward a Brighter Future by Siegfried M. Pueschel, 1990

Party Train by Lesli Mitchell and illustrated by Ramon Gil, 2001 (Written for children with Developmental Disabilities)

Pediatric Nutrition in Chronic Diseases and Developmental Disorders: Prevention, Assessment, and Treatment edited by Shirley Walberg Ekvall, 1993

Pediatric Traumatic Brain Injury: Proactive Intervention by Jean Blosser, 2003

Person-Centered Planning and Outcome Management: Maximizing Organizational Effectiveness in Supporting Quality Lifestyles Among People with Disabilities by Jane M. Everson and Dennis H. Reid, 1999

“Phenylketonuria” videotape produced by Films for the Humanities & Sciences, 2001

The PKU Cookbook by Elizabeth Read, et al., 1976

Plain Talk on Developmental Disabilities: A Guide for Service Providers by Mac Griffith, 1998

Play and Imagination in Children with Autism by Pamela J. Wolfberg, 1999

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A Practical Guide to Respite for Your Family by Molly Dellinger-Wray and Monica Uhl, 1996

Pretending to Be Normal: Living with Asperger's Syndrome by Liane Holiday Willey, 1999

Primary Care of the Child with a Chronic Condition edited by Patricia Ludder Jackson and Judith A Vessey, 2000

Raising a Child with Autism: A Guide to Applied Behavior Analysis for Parents by Shira Richman, 2001

Retarded Isn't Stupid, Mom! by Sandra Z. Kaufman, 1999 (Mental Retardation)

"Rett Syndrome: A Closer Look" videotape produced by the International Rett Syndrome Association, 1992

Right Address . . . Wrong Planet: Children with Asperger Syndrome Becoming Adults by Gena P. Barnhill, 2002

Ryan - A Mother's Story of Her Hyperactive/Tourette Syndrome Child by Susan Hughes, 1990

"Sean's Story: A Lesson in Life" videotape produced by Files for the Humanities & Sciences, 1994 (Down Syndrome)

"Silent Angels: The Rett Syndrome Story" videotape produced by International Rett Syndrome Association, 2000

Solving Behavior Problems in Autism: Improving Communication with Visual Strategies by Linda A. Hodgdon, 1999

SPINAbilities: A Young Person's Guide to Spina Bifida edited by Marlene Lutkenhoff and Sonya G. Oppenheimer, 1997

"Students Like Me: Teaching Children with FAS" videotape produced by Betsy Anderson, 2000

Taking the Mystery Out of Medications in Autism/Asperger Syndromes: A Guide for Parents and Non-Medical Professionals by Luke Y. Tsai, 2001

Targeting Autism: What We Know, Don't Know, and Can Do to Help Young Children with Autism and Related Disorders by Shirley Cohen, 1998

Teaching Reading to Children with Down Syndrome: A Guide for Parents and Teachers by Patricia Logan Oelwein, 1995

This is Asperger Syndrome by Elisa Gagnon and Brenda Smith Myles, 1999

A Thousand Words: Healing Through Art for People with Developmental Disabilities by Cindy Caprio-Orsini, 1996

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Thyroid Diseases of Infancy and Childhood: Effects on Behavior and Intellectual Development edited by Peter Hauser and Joanne Rovet, 1999

Toilet Training for Individuals with Autism & Related Disorders: A Comprehensive Guide for Parents & Teachers by Maria Wheeler, 1998

Traumatic Head Injury in Children edited by Sarah H. Broman and Mary Ellen Michel, 1995

“Traumatic Brain Injury” videotape presented by Reed Martin, J.D., 1992

Understanding the Occurrence of Secondary Disabilities in Clients with Fetal Alcohol Syndrome (FAS) and Fetal Alcohol Effects (FAE): Final Report by Ann Streissguth, et al., 1996

Visual Recipes: A Cookbook for Non-Readers by Tabitha Orth, 2000

“Waddie Welcome: A Man Who Cannot be Denied” videotape produced by Narcel G. Reedus and University of Georgia (Cerebral Palsy)

We Can Do It! by Laura Dwight, 1992 (Spina Bifida, Down Syndrome, Cerebral Palsy)

What is Asperger Syndrome and How Will it Affect Me? by Martine Ives and illustrated by Matt Watkins, 2001

When Autism Strikes: Families Cope with Childhood Disintegrative Disorder edited by Robert A Cartalano, 1998

Wild Child: How You Can Help Your Child with Attention Deficit Disorder (ADD) and Other Behavioral Disorders by Don Mordasini, 2001

“Writing Social Stories with Carol Gray” videotape produced by Future Horizons, 2000 (Autism)

“Worth the Trip: Raising and Teaching Children with Fetal Alcohol Syndrome” videotape produced by Vida Health Communications, 1996

Your Child has a Disability: A Complete Sourcebook of Daily and Medical Care by Mark L. Batshaw, 1998

Your Values, My Values: Multicultural Services in Developmental Disabilities by Lilah Morton Pengra, 2000

# GLOSSARY

**active range of motion:** Independent movement of body parts through an arc.

**activities of daily living (ADL):** Include the following: grooming, oral hygiene, bathing, toilet hygiene, dressing, feeding/eating, medication routine, socialization, functional communication, functional mobility, sexual expression.

**Americans with Disabilities Act (ADA):** Federal legislation that gives civil rights protection to individuals with disabilities. Enacted into law July, 1990.

**amniocentesis:** A prenatal assessment of a fetus which involves analysis of amniotic fluid.

**amniotic fluid:** Fluid that surrounds and protects the developing fetus. This fluid is sampled through amniocentesis

**anxiety:** Apprehension, tension or uneasiness that stems from the anticipation of danger which may be internal or external. Some definitions of anxiety distinguish it from fear by limiting it to the anticipation of danger from a largely unknown, source, whereas fear is the response to a consciously recognized and usually external threat or danger. The manifestations of anxiety and fear are not the same and include motor tension, autonomic hyperactivity, apprehensive expectation and vigilance and scanning.

**atonia:** A condition evidenced by lack of muscle tone.

**atrophy:** A wasting away.

**blind:** A person either totally blind or having very poor eyesight. More specifically, having no vision (with the use of eye glasses) better than 20/200 in the better eye, or a limited visual field of 20 degrees or less.

**caregivers:** Any persons who have input into the care of the child: babysitter, extended family, day care workers, hospital workers (nurses, aides, etc.).

**chromosomal abnormalities:** Defects or damage in the chromosomes of an individual.

**chromosomes:** Threadlike materials within a cell that carry the genes; and therefore, play a central role in tissue development and inherited characteristics.

**chronic:** Marked by long duration or frequent recurrence.

**clonus:** Involuntary, rapid contractions and relaxations of a muscle.

**cognitive:** A term that describes the process people use for remembering, reasoning, understanding, problem solving, evaluating, and using judgment. Cognition more simply, is what a person or child knows and understands, or the process of knowing.

**cognitive development:** The development of skills necessary for understanding and organizing the world, including such perceptual and conceptual skills as discrimination, memory, sequencing, concept formation, generalization, reasoning, and problem solving.

**cognitive functioning:** Refers to the level of proficiency in thinking, processing information, and knowledge.

# GLOSSARY

**communication skills:** Consciously linking the meaning and the purpose of what (we say) is said to what is (we do) done.

**congenital:** Present at birth. A condition or disease existing at birth, that is not necessarily caused by inheritance.

**contraction:** Shortening of a muscle to cause movement.

**contracture:** Stiffening or shortening of muscle caused physiologically and/or by lack of use; result in a reduction of the range of movement of a joint. For example, if an elbow or knee remained bent for extended periods, it could become more and more difficult to straighten.

**coordination:** Acting together in a smooth way. Several muscle groups working together in harmony.

**cornea:** The external covering of the eye.

**deaf:** 1. A term used to categorize individuals who have hearing losses greater than 75 to 80 dB, have vision as their primary input, and cannot understand speech through the ear even with the use of hearing aids. The sense of hearing for a person who is deaf is nonfunctional for the ordinary purposes of life. 2. As defined in P.L. 94-142: Hearing impairment so severe as to impede the child from processing linguistic information through hearing, with or without amplification, and which adversely affects educational performance.

**development:** Growing both physically and mentally.

**developmental:** Having to do with the steps or stages in growth and development before the age of 18.

**developmental delay:** When a child's development progresses at a slower rate than most children.

**developmental disability (DD):** 1. A handicap or impairment originating before the age of 18 which may be expected to continue indefinitely and which constitutes a substantial impairment. The disability may be attributable to mental retardation, cerebral palsy, epilepsy, or other neurologic conditions and may include autism. 2. According to the Developmental Disabilities Act: When applied to infants and young children it means: Individuals from birth to age 5, inclusive, who have substantial developmental delay or specific congenital or acquired conditions with a high probability of resulting in developmental disabilities if services are not provided. For persons 5 years of age or older it's defined as: A severe, chronic disability which: (A) is attributable to a mental or physical impairment or combination of mental and physical impairments; (B) is manifested before the person attains age twenty-two; (C) is likely to continue indefinitely; (D) results in substantial functional limitations in three or more of the following areas of major life activity: (i) self care, (ii) receptive and expressive language, (iii) learning, (iv) mobility, (v) self-direction, (vi) capacity for independent living, and (vii) economic self-sufficiency; and (E) reflects the person's need for a combination and sequence of special, interdisciplinary, or generic care, treatment, or other services which are of lifelong or extended duration and are individually planned and coordinated.

**diagnosis:** 1. Naming the cause of a disorder by looking at its symptoms. 2. The process of identifying specific mental or physical disorders. Some use the term more broadly to refer to a comprehensive evaluation not limited to the identification of specific disorders.



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**epicanthal fold:** A vertical fold of skin on either side of the nose, which covers the innermost portion of the eye. The presence of this fold gives individuals with Down Syndrome the appearance of slanted eyes.

**expressive language:** The ideas, concepts and feelings the child is able to share through speech, signing, gestures, etc.

**eye contact:** "Looking him in the eye" while talking to the listener; generally a natural, although not a constant, interaction of the speaker's eyes with those of the listener. Varies according to a person's culture.

**failure to thrive (FTT):** A chronic disorder of infancy and childhood characterized by growth failure, malnutrition and variable degrees of the delay in motor and social development. Possible causes of FTT are varied; illness, oral-motor feeding and swallowing disorders, inadequate food resources and problems with parent-child interaction.

**fine motor:** The use of small muscle groups for controlled movements, particularly in object manipulation. Such as movements our hands make, how we hold onto things, move our fingers, etc.

**gait:** A particular pattern or style in which a person walks.

**genetic counseling:** A process of informing parents concerning decisions they have to make regarding having children; often done where there is some reason to believe that a genetic abnormality may result, or be present.

**genetic screening:** A search in a population for persons possessing certain genotypes (genes transmitted from parents to offspring) that are (1) already associated with disease or predisposed to disease, (2) may lead to disease in their descendants, or (3) produce other variations not known to be associated with disease.

**geneticist:** A person who studies the hereditary characteristics of families. Also referred to as a genetic specialist.

**gross motor:** Movement that involves balance, coordination and large muscle activity as required in holding your head up, walking, running, skipping, climbing, jumping and other physical activities.

**hormone:** An internally secreted compound formed in endocrine organs.

**hypertelorism:** Widely spaced eyes.

**hypertonia:** High muscle tone (stiff muscles).

**hypotonia:** 1. Low, or poor muscle tone (floppy muscles). 2. Damage to any part of the brain, usually including the cerebellum and basal ganglia, resulting in decreased stiffness of the extremities and trunk.

**impairment:** Something that someone lacks - it can be mental, visual, hearing, or weakness in an arm. An impairment can either be acquired during the course of a lifetime, or congenital (born with).

**infantile:** Relating to or characteristic of infants or infancy.

**intelligence quotient (IQ):** A score obtained from an intelligence test that provides a measure of mental ability in relation to age.

# GLOSSARY

**juvenile:** Pertaining to or denoting youth, childhood, immaturity.

**language delay:** A term used when the normal rate of language development is interrupted, but the developmental sequence remains intact.

**low birth weight:** A term applied to babies that weigh 5 1/2 pounds (2,500 grams) or less at birth. Low birth weight infants are of two different types: those who are born too small because they are born too soon, and those who are born on time, but are too small for their gestational age.

**macrocephaly:** An abnormally large head.

**microencephaly:** A condition in which the head and brain are significantly smaller than normal for age and sex (head circumference less than the 5th percentile for age). May be associated with mental retardation.

**muscle tone:** Amount of tension in a muscle at rest.

**mutation:** A change in the genetic material that occurs by chance.

**neonatal:** The first four weeks after a child's birth.

**perception:** A person's ability to consciously recognize and interpret what is seen, heard, or felt. More specifically, the process of organizing or interpreting the raw data (stimuli) obtained through the senses.

**physical development:** Growth. Biogenetically based changes in a child's physical characteristics, including changes in weight, height, skeletal and muscular features, and maturation of the circulatory, respiratory and nervous systems.

**pigmentation:** Coloration of the skin and eyes.

**prenatal:** The time before birth, while a baby is developing during pregnancy. The period of time between the conception and birth of an infant.

**progressive:** A gradual worsening.

**range of motion:** How far you can bend your body parts. More specifically, the range measured in degrees of a circle through which a joint can be moved.

**receptive language:** Language that is spoken or written by others and received by an individual. The receptive language skills are listening and reading.

**sensory:** Relating to the various sensory systems: tactile, kinesthetic, olfactory, visual, auditory, gustatory, vestibular.

**sensory stimulation:** Provide input to the different sensory systems to be received, differentiated and interpreted.

**short stature:** Height/length less than the 5th percentile for age.

**social skills:** Skills related to social interactions with peers.

# GLOSSARY

**spasticity:** Increased muscle tone (hypertonic), involuntary resistance of weak muscle caused by passive range of motion followed by sudden relaxation of muscle, associated with exaggeration of reflexes. Causes stiffness, awkward movements, and loss of voluntary muscle control.

**speech:** The mechanical production of sounds and words through the voice.

**speech and language disorders:** Difficulties in communicating effectively.

**stature:** Height. Distance from the crown of the head to the bottom of the feet when the person is standing.

**sucking:** A rhythmical method for obtaining liquid and food characterized by the active coordination of small up/down jaw movements, up/down tongue movements, lip approximation, and cheek activity; negative pressure is built up in the oral cavity due to the more closed mouth position.

**syndrome:** A combination of symptoms which occur together and define a disease or disorder.

**term infant:** An infant born between 38 and 40 weeks gestation, some may consider 37 weeks term.

The definitions in this glossary were taken from the Dictionary: For Parents of Children with Disabilities. The complete Dictionary: For Parents of Children with Disabilities is available on the Center for Disabilities website at [www.usd.edu/cd](http://www.usd.edu/cd).

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