

Guide to developmental Disorders

Part 2

Genetic and Other Disorders

Introduction

Here are some rare and not so rare genetic Disorders, and some disorders caused by injury to the brain. These injuries can be caused before, during or after birth of the child .

This is just an indicative list with only the basic information. This is ONLY for initial reference.

This mentions the name, what it is, symptoms, diagnosis, medical treatment or therapies, and prognosis.

A few useful links are given for each disorder . There is a lot of information available both scientific and medical which can be found under these and also more links to other sites.

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1Angelman Syndrome (AS)

What is AS?

Angelman Syndrome (AS) is a rare neuro-genetic disorder named after an English pediatrician, Dr. Harry Angelman, who first described the syndrome in 1965.

A syndrome is number of features which occur together as a group and indicate a particular condition.

AS is characterized by severe intellectual disability, speech impediment, sleep disturbance, unstable jerky gait, seizures and usually a happy demeanor.

Symptoms are usually evident after the age of three, and are characterized by severe congenital mental retardation, unusual facial appearance, and muscular abnormalities.

Causes

Researchers have found a very small deleted area in chromosome 15 in patients with Angelman Syndrome. The Angelman Syndrome gene is called UBE3A.

There is no known way to prevent Angelman Syndrome.

Symptoms

Symptoms of Angelman syndrome include a stiff, unstable jerky gait, absent or diminished speech skills, hand flapping, excessive laughter/unusually happy demeanor, developmental delay, and small head size (microcephaly). Some patients may also develop epilepsy and have problems with balance.

Laughter is very frequent in AS. Studies of the brain in AS, using MRI or CT scans, have not shown any defect suggesting a site for a laughter-inducing abnormality. The laughter in AS seems mostly to be an expressive motor event.. Although AS children experience a variety of emotions, apparent happiness predominates.

The first evidence of this distinctive behavior may be the onset of early or persistent social smiling at the age of 1-3 months. Giggling, chortling and constant smiling soon develop and appear to represent normal reflexive laughter but cooing and babbling are delayed or reduced.

Diagnosis

Because it is difficult to detect the developmental problems associated with Angelman syndrome during infancy, children are usually diagnosed with the disorder between the ages of three and seven. Parents and doctors may notice a

developmental delay between the ages of 6 and 12 months, but because the child does reach most developmental milestones during this stage, a diagnosis cannot be made. Brain scans are also normal at this time.

By the age of two, however, the child with Angelman Syndrome will begin to show signs of microcephaly. By age three, clinical features of Angelman Syndrome are present, including speech impairment, movement or balance disorder, frequent laughter and smiling, easily excitable personality, hand flapping movements, and short attention span.

Seizures may begin after the age of three. An abnormal electroencephalograph (EEG), a painless procedure in which wires are pasted to the scalp to record the brain's electrical activity, will also be found at this time.

Treatment

Till date there is no cure for AS. The condition is permanent but is not degenerative. As such children can look forward to a normal lifespan.

Treatment of Angelman Syndrome is focused on providing physical therapy and adaptive devices to assist with gait and balance problems. Speech therapy is recommended for language problems. Patients with epilepsy are prescribed anticonvulsant medications.

Many educational and behavioral interventions are effective in addition to physical and occupational therapies, speech and language interventions, behavior modification, and parent training.

For more information, please visit:

Angelman Syndrome Foundation. <http://www.angelman.org/>

Geneclinics (Developed at the University of Washington, Seattle)

<http://www.geneclinics.org/profiles/angelman/details.html>

Angelman Syndrome – facts for Families <http://www.asclepius.com/angel/>

Penn State Children's Hospital

<http://www.hmc.psu.edu/childrens/healthinfo/a/angelman.htm>

<http://asclepius.com/angel/asfinfo.html#intro>

Angelman Syndrome Association <http://www.angelmansyndrome.org/home.html>

Here you can find international organizations' list

<http://www.angelmansyndrome.org/links.html>

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2. Bipolar Disorder

What is Bipolar Disorder

Bipolar disorder is a serious medical illness that can affect a person's ability to feel a normal range of moods. People with bipolar disorder have mood swings that can range from very low (depression) to very high (mania).

Bipolar disorder is also known as manic depression. The word "bipolar" is now used because the disorder is made up of two poles, or extremes.

Bipolar disorder may cause behavior so severe that you may not be able to function at work, in family or social situations, or in relationships with others. Some people with bipolar disorder become suicidal.

People with extreme cases of mania may be hospitalized to protect them from risky behaviors, such as driving recklessly or having delusions. During a severe bout of depression, hospitalization may be necessary to prevent the person from committing suicide.

Though it is a Mental Illness it is NOT Madness.

Causes

The illness tends to be highly genetic, but there are clearly environmental factors also that influence whether the illness will occur in a particular child.

An imbalance of chemicals in the brain is another possible cause.

Bipolar disorder can skip generations and take different forms in different individuals

Symptoms

The mood changes of bipolar disorder can be mild or extreme. They may develop gradually over several days or weeks, or come on suddenly within minutes or

hours. The manic or depressive episodes may only last a few hours or for several months.

Types of Episodes

According to the American Psychiatric Association, bipolar disorder includes the following four types of mood episodes:

- **Depression** - People with bipolar disorder can feel very sad, sometimes for long periods of time. They may not even want to get out of bed or eat. They don't enjoy doing things they used to do.
- **Mania** - Mania is the other side of bipolar disorder. Mania may start with a good feeling, almost like a "high." Or it may make a person feel very irritable and angry. People with mania may do very risky things.
- **Hypomania** - Hypomania is a milder form of mania. A person may feel good and may think that he or she is getting more things done. But the "feel good" stage can change into full-blown mania or depression.
- **Mixed mood** - Feelings of mania and depression can also go back and forth in the same day. This is called a mixed episode.

With all types of episodes, patients are at risk for suicide

Mania Symptoms:

- Increased energy level
- Less need for sleep
- Racing thoughts or mind jumps around
- Easily distracted
- More talkative than usual or feeling pressure to keep talking
- More self-confident than usual
- Focused on getting things done, but often completing little
- Risky or unusual activities to the extreme, even if it's likely bad things will happen

Depression Symptoms:

- Feeling sad or blue, or "down in the dumps"
- Loss of interest in things the person used to enjoy, including sex
- Feeling worthless, hopeless, or guilty
- Sleeping too little or too much
- Changes in weight or appetite
- Feeling tired or having little or no energy
- Feeling restless
- Problems concentrating or making decisions
- Thoughts of death or suicide

Diagnosis

Because it has many phases and symptoms, bipolar disorder is complex and hard to diagnose. There are no lab tests for bipolar disorder; instead your doctor or therapist will ask detailed questions about what kind of symptoms you have and how long they last. In order to be diagnosed as bipolar, you must have had a manic episode lasting at least a week (less if you had to be hospitalized). During this time, you must have had three or more specific symptoms of mania, such as needing less sleep, being more talkative, behaving wildly or irresponsibly in activities that could have serious outcomes, or feeling as if your thoughts are racing.

Your urine and blood may be tested to rule out other problems that could be causing your symptoms.

Treatment

Bipolar disorder is a lifelong illness. But today there are many treatments for people with bipolar disorder.

The first medical breakthrough in the treatment of bipolar disorder came in the 1950s. Treatment has made great strides since then. Today, there are many medications and therapies used to manage bipolar disorder. People with bipolar disorder can hold jobs, raise families, and live normal lives.

There are three basic parts of treatment for bipolar disorder. They are medication, therapy, and self-care.

1. Medication

These medications may be used alone or with others. Some medications may take time to start working. Types of medications for bipolar disorder include the following:

- **Mood stabilizers** may delay or relieve episodes of mania (highs) or depression (lows).
- **Antidepressants** are often used with a mood stabilizer in people with depressive episodes.
- **Antipsychotics** are medications that are mostly used to treat mania , severe anxiety, sleeplessness extreme nervousness or agitation.

Electroconvulsive therapy (ECT), although not a medication, is a medical treatment used to help people with severe mania or depression. It can also help people who do not do well with medication.

2. Talk Therapy

Talk therapy is also called psychotherapy. It is talking with a trained Psychologist about yourself, your situation, relationships, and condition. It can help you learn how to make sense of your thoughts and feelings.

There are different settings for psychotherapy—some are one-on-one and some are in a group setting. Some types provide education and support. Others focus on learning about yourself or your relationships with others.

3. Self-Care

Taking an active role in managing bipolar disorder is important. Here are some things you can do to help yourself:

- Read and learn about bipolar disorder.
- Try to learn and avoid triggers of mood episodes.
- Take medication as prescribed
- Do some de-stressing exercises regularly
- Avoid Alcohol and Drugs
- Join a support Group.

Prognosis

Living with bipolar disorder is much like living with other medical illnesses. With proper medication, education, and support, bipolar disorder can usually be treated or managed effectively. Therapy, support groups, coaching, family, and friends can help.

It's like having an illness such as diabetes or heart disease. People with bipolar disorder must manage their condition carefully.

The day-to-day job of managing bipolar disorder belongs to you. But that does not mean you are alone. Consider asking for help and support from your trusted family and friends.

For more information, please visit:

Child and Adolescent Bipolar Foundation <http://www.bpkids.org/site/PageServer>

Yahoo <http://health.yahoo.com/ency/healthwise/hw148751>

Bipolar.com <http://www.bipolar.com/>

List of Resources <http://www.bipolar.com/resources.html>

Yahoo <http://health.yahoo.com/ency/healthwise/hw148751>

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3.Down Syndrome

What is it ?

Down syndrome is the most common and readily identifiable chromosomal condition associated with mental retardation. It is caused by a chromosomal abnormality: for some unexplained reason, an accident in cell development results in 47 instead of the usual 46 chromosomes. This extra chromosome changes the orderly development of the body and brain. In most cases, the diagnosis of Down syndrome is made according to results from a chromosome test administered shortly after birth.

It is named after John Langdon Down, an English physician who published an accurate description of a person with Down syndrome.

Cause

We don't know what causes the extra 21st chromosome. We know that the extra 21st chromosome causes an extra dose of some proteins. Those proteins cause the typical features of Down Syndrome. We don't know most of the proteins involved and how they cause Down Syndrome. We do know there is an association between a mother's age and the chances of having a baby with Down Syndrome. No causes have been identified and proven, however.

Symptoms

There are over 50 clinical signs of Down syndrome, but it is rare to find all or even most of them in one person. Some common characteristics include:

- Mongoloid features
- Hyperflexibility (excessive ability to extend the joints);
- Short, broad hands with a single crease across the palm on one or both hands;
- Broad feet with short toes;
- Flat bridge of the nose;
- Short, low-set ears;
- Short neck;

- Small head;
- Small oral cavity; and/or
- Short, high-pitched cries in infancy.

Individuals with Down syndrome are usually smaller than their non-disabled peers, and their physical as well as intellectual development is slower.

Diagnosis

The diagnosis is usually suspected at birth due to the presence of physical characteristics such as a large tongue, heart problems, poor muscle tone, and flat facial features. It is confirmed through chromosomal testing.

Prognosis

The genetic make-up cannot be changed . It is a lifelong condition. The effects of Down Syndrome, however, can be modified by providing good medical care, good education and good parental support.

Medical treatments

Although there have been proposed medical treatments for Down Syndrome, none of them have been proven effective.

For more information, please visit:

[National Down Syndrome Society](http://www.ndss.org/)

<http://www.ndss.org/>

For up-to-date list of world organizations:

<http://downsyndrome.com>

National Down Syndrome Congress
1370 Center Drive, Suite 102
Atlanta, GA 30338
800.232.6372; 770.604.9500
E-mail: info@ndscenter.org
Web address: www.ndscenter.org

National Down Syndrome Society
666 Broadway
New York, NY 10012
212.460.9330
800.221.4602 (Toll Free)
E-mail: info@ndss.org
Web address: www.ndss.org

The Arc of the United States (formerly the Association for Retarded Citizens of the

United States)
1010 Wayne Avenue, Suite 650
Silver Spring, MD 20910
301.565.3842
E-mail: info@thearc.org
Web address: www.thearc.org

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4.Fragile X Syndrome

What is Fragile X Syndrome ?

Fragile X is a family of genetic conditions, which can impact individuals and families in various ways. These genetic conditions are related in that they are all caused by gene changes in the same gene, called the FMR1 gene.

Fragile X Syndrome is the most common form of inherited mental retardation and the most common known cause of autism.

It is more common in boys than girls. Individuals with this disorder often have distinctive physical features, such as a long face, large prominent ears and hyperextensible joints. DNA testing can determine a diagnosis for Fragile X, which is a lifelong disorder with no cure, but treatable with behavioral and educational therapies.

Symptoms

Fragile X syndrome affects individuals in a wide variety of ways. Some individuals experience significant challenges because of the effects of fragile X, while the impact on others is so minor that they will never be diagnosed.

Males and females exhibit quite different physical, cognitive, behavioral, sensory, speech and language impacts of fragile X syndrome. There may be problems with the sexual organs.

In general, females with fragile X either do not have the characteristics seen in males, or the characteristics show up in a milder form.

Males

The primary physical features that people associate with fragile X syndrome in males are long faces and prominent ears. These features are more common in males over the age of 10 than among those under 10.

The ears are often wider and longer than usual and may project away from the head. However, long ears are also common among mentally retarded males who do not have fragile X syndrome.

When compared to mentally retarded males who do not have fragile X syndrome, those with fragile X have a larger head circumference, head breadth and head length.

Females

A premutation in the FMR1 gene typically has little or no impact on a female in terms of behavior and educational ability. However several facial characteristics such as prominent ears and prominent jaw appear commonly in females with the premutation.

Diagnosis

Fragile X syndrome is difficult to diagnose in children. An affected infant may develop normally at first. After age one year, though, the child begins to have noticeable delays in language and short-term memory. Physical signs of Fragile X, such as the typical facial features, are only noticeable after the onset of puberty.

Fragile X syndrome may be suspected if the individual has a number of male relatives with mental retardation. However, in many families the child with Fragile X syndrome is the first member known to have mental retardation. If Fragile X syndrome is suspected, a blood test known as FMR1/DNA is used to confirm the diagnosis.

Treatment

Medical care for an individual with Fragile X syndrome focuses on treating common problems and helping the individual achieve his/her developmental potential. Screenings for scoliosis, heart valve defects, and vision problems will be performed.

Prognosis

Children affected by Fragile X will benefit from educational support services and behavior management. Fragile X does not shorten a person's life span.

For more information, please visit:

FRAXA Research Foundation	http://www.fraxa.org/
Family Village	http://www.familyvillage.wisc.edu/lib_frgx.htm
The National Fragile X Foundation	http://www.nfxf.org/
The Fragile X society	http://www.fragilex.org.uk/
<u>More links from here</u>	http://www.nfxf.org/html/other_links.htm

Fragile X Society

Road End House
6 Stortford Road
Great Dunmow, Essex, CM6 1DA
UK
Tel: 01371 875100
Fax: 01371 859915
Email: info@fragilex.org.uk
Internet: <http://www.fragilex.org.uk>

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5. IsoDicentric 15

What is isodicentric 15?

Isodicentric 15, abbreviated idic(15), is one of a group of genetic conditions.

Most children and adults with idic(15) experience developmental disabilities. These can range from mild to severe and may be accompanied by other neurological, physical, and behavioral problems.

Cause

Isodicentric chromosome 15 is the scientific name for a specific type of chromosome abnormality. Individuals with isodicentric chromosome 15, or "idic(15)", have 47 chromosomes instead of the typical 46 chromosomes.

Individuals with idic(15) usually have a total of four copies of this chromosome 15 region instead of the typical two copies (1 copy each on the maternal and paternal chromosomes and 2 copies on the idic(15)). It is not known why this happens.

Symptoms

Most children with idic(15) do not appear unusual, although many share similar facial characteristics. These include a flat nasal bridge which gives them a 'button' nose. There may be skin folds, called 'epicanthi', at the inner corners of the eyes. The palate (roof of the mouth) may be unusually high. Rarely, babies with idic(15) may be born with a cleft lip and/or palate or differences in the way their hearts, kidneys, or other body organs are formed.

These people have a tendency for seizures.

Diagnosis

The extra chromosome in people with idic(15) can be easily detected through a blood test called a chromosome study. An additional genetic test, called FISH (Fluorescence In Situ Hybridization), confirms the diagnosis by distinguishing idic(15) from other supernumerary marker chromosomes.

Treatment

At the present time, there is no specific treatment which can undo the genetic pattern seen in people with idic(15). The extra chromosomal material in those affected was present at or shortly after conception, and its effects on brain development began taking place long before the child was born.

Although the fundamental genetic difference cannot be reversed, therapies are available to help address many of the symptoms associated with idic(15). Medication for seizures is available, and physical, occupational, and speech therapies for other developmental problems.

For more information, please visit:

I.D.E.A.S. IsoDicentric 15 Exchange, Advocacy & Support

<http://www.idic15.org/>

Contact A Family

<http://www.cafamily.org.uk/Direct/i17.html>

First signs

http://www.firstsigns.org/delays_disorders/other_disorders.htm

Birth Disorder Information Directory

<http://www.bdid.com/defecti.htm>

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6.Mental Retardation

What is it ?

Mental retardation (MR) is a developmental disability that first appears in children under the age of 18. It is defined as a level of intellectual functioning (as measured by standard intelligence tests) that is well below average and results in significant limitations in the person's daily living skills (adaptive functioning).

A person with mental retardation may have difficulties with communication, conceptual skills, social skills, self-care, home living, social skills, community use, self-direction, health and safety, functional academics, leisure, and work.

These people have an IQ below average (<70).

Cause

A variety of problems can lead to mental retardation. The causes of mental retardation can be divided into Four broad classifications, - genetic factors, prenatal illnesses and exposures, childhood illnesses and injuries, and environmental factors.

Genetic Factors

There are now over 750 known genetic disorders that cause mental retardation. Some of these can be screened for during pregnancy—including Down syndrome, the most common chromosomal cause of mental retardation.

Other disorders include fragile X syndrome, Prader-Willi syndrome, and Williams syndrome. People with these and other syndromes often show distinctive personalities, behavioral problems, and intellectual strengths and weaknesses that can be used to guide their care.

Prenatal Illnesses and Exposures

Fetal alcohol syndrome results from the mother's drinking during the first 12 weeks (1st trimester) of pregnancy. Drug abuse and cigarette smoking during pregnancy have also been linked to mental retardation. It is generally accepted that pregnant women should avoid all alcohol, tobacco, and recreational drugs.

Maternal infections and such illnesses as glandular disorders, rubella, toxoplasmosis, and cytomegalovirus (CMV) infection may cause mental retardation. When the mother has high blood pressure (hypertension) or blood poisoning (toxemia), the flow of oxygen to the fetus may be reduced, causing brain damage and mental retardation.

Birth defects that cause physical deformities of the head, brain, and central nervous system frequently cause mental retardation.

Childhood Illnesses and Injuries

Hyperthyroidism, whooping cough, chicken pox, measles, and Hib disease (a bacterial infection) may cause mental retardation if they are not treated adequately. An infection of the membrane covering the brain (meningitis) or an inflammation of the brain itself (encephalitis) can cause swelling that in turn may cause brain damage and mental retardation. Traumatic brain injury caused by a blow to the head or by violent shaking of the upper body may also cause brain damage and mental retardation in children.

Environmental Factors

Ignored or neglected infants who are not provided with the mental and physical stimulation required for normal development may suffer irreversible learning impairment. Children who live in poverty and suffer from malnutrition, unhealthy living conditions, abuse, and improper or inadequate medical care are at a higher risk.

Exposure to lead or mercury can also cause mental retardation. Many children have developed lead poisoning from eating the flaking lead-based paint often found in older buildings.

Types of MR

Mental Retardation is usually classified by the child's IQ:

- **Mild:** IQ of 55-69. About 85% of children with mental retardation are in this range and are further classified as being **educable**. These children may be able to learn to read and write at the 4th or 5th grade level, live relatively independently and work with special training.
- **Moderate:** IQ of 40-54. Also described as being **trainable**. 10% of children with mental retardation are in this group. These children may have academic potential at the kindergarten or 1st grade level and may have limited ability to read, and will usually need some support and supervision in daily living activities (like with a supportive family or supervised group home) and work (with special training).
- **Severe:** IQ of 25-39. 5% of children with mental retardation are in this group. Children with this level of mental retardation are unlikely to be able to learn to read or write, but may be able to be toilet trained and learn to dress with assistance. They usually require total supervision and support for daily living activities.
- **Profound:** IQ of < 24. <1% of children with mental retardation are in this group. They are totally dependent on outside support.

A newer classification system was developed in 1992 that is based not on IQ scores, but rather on the amount of support and supervision that the individual needs: intermittent, limited, extensive and pervasive.

Diagnosis

If mental retardation is suspected, a comprehensive physical examination and medical history should be done immediately to discover any organic cause of symptoms.

Such conditions as hyperthyroidism and PKU are treatable. The progression of retardation can be stopped and, in some cases, partially reversed if these conditions are discovered early.

If a neurological cause such as brain injury is suspected, the child may be referred to a neurologist or neuropsychologist for testing.

Genetic Testing is done through chromosomal analysis for Down Syndrome or Fragile X Syndrome, or more extensive testing may be done, including an MRI of the brain and metabolic screens for others.

Testing will also include **psychological tests** to evaluate their IQ and level of functioning

Treatment

Though there is not a cure available for Retardation, training in independent living and job skills is often begun in early adulthood. The level of training depends on the degree of retardation. Mildly retarded people can often acquire the skills needed to live independently and hold an outside job. Moderate to profoundly retarded persons usually require supervised community living in a group home or other residential setting.

Prognosis

People with mild to moderate mental retardation are frequently able to achieve some self-sufficiency and to lead happy and fulfilling lives. To reach these goals, they need appropriate and consistent educational, community, social, family, and vocational supports.

The outlook is less promising for those with severe to profound retardation. Studies have shown that these persons have a shortened life expectancy. The diseases that are usually associated with severe retardation may cause the shorter lifespan. People with Down syndrome will develop the brain changes that characterize **Alzheimer's disease** in later life and may develop the clinical symptoms of this disease as well.

Prevention

Immunization against diseases such as measles and Hib prevents many of the illnesses that can cause mental retardation. In addition, all children should undergo routine developmental screening as part of their pediatric care.

Newborn screening and immediate treatment for PKU and hyperthyroidism can usually catch these disorders early enough to prevent retardation.

Good prenatal care can also help prevent retardation. Pregnant women should be educated about the risks of alcohol consumption and the need to maintain good nutrition during pregnancy. Such tests as amniocentesis and ultrasonography can determine whether a fetus is developing normally in the womb.

For more information, please visit:

The ARC and the American Association on Mental Retardation (AAMR)

<http://www.aamr.org/>

Keep Kids Healthy.com

http://www.keepkidshealthy.com/welcome/conditions/mental_retardation.html

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7. Obsessive Compulsive Disorders (OCD)

What is OCD?

OCD is a medical brain disorder that causes problems in information processing. It is not your fault or the result of a "weak" or unstable personality.

Obsessive-Compulsive Disorder (OCD) is a serious anxiety-related condition that affects as many as three in a hundred people – from young children to older adults - regardless of gender and social or cultural background.

People with OCD worry a **lot**. And they feel afraid about bad things that could possibly happen. To some degree OCD-type symptoms are probably experienced at one time or another by most people, especially in times of stress. However, the illness can have a totally devastating effect on work, social life and personal relationships. The World Health Organisation (WHO) even ranks OCD as the tenth most disabling illness of any kind, in terms of lost earnings and diminished quality of life.

These frequent worry thoughts are called obsessions, and the behaviors people do to try to make the worry thoughts go away are called compulsions.

Causes

Obsessive-compulsive disorder isn't contagious, so you can't catch it from someone like you can a cold.

There is no proven cause of OCD. Research suggests that OCD involves problems in communication between the front part of the brain (the orbital cortex) and deeper structures (the basal ganglia). These brain structures use the chemical messenger serotonin. It is believed that insufficient levels of serotonin are involved in OCD. Drugs that increase the brain concentration of serotonin often help improve OCD symptoms.

No specific genes for OCD have been identified. Research suggests that genes do play a role in the development of the disorder. Childhood-onset OCD runs in families.

OCD starts at any time from preschool age to adulthood (usually by age 40). One third to one half of adults with OCD report that it started during childhood.

Symptoms

OCD involves having both obsessions and compulsions. A person with OCD may sometimes have one or the other

1. Obsessions

Obsessions are thoughts, images, or impulses that occur over and over again and feel out of your control. The person does not want to have these ideas. He finds them disturbing and intrusive, and usually recognizes that they don't really make sense. People with OCD worry excessively about dirt and germs and become obsessed with the idea that they are contaminated or contaminate others. Obsessions are accompanied by uncomfortable feelings, such as fear, disgust, doubt, or a sensation that things have to be done in a way that is "just so."

2. Compulsions

People with OCD try to make their obsessions go away by performing compulsions. Compulsions are acts the person performs over and over again, often according to certain "rules." People with an obsession about contamination may wash constantly to the point that their hands become raw and inflamed. A person may repeatedly check that she has turned off the stove or iron because of an obsessive fear of burning the house down. She may have to count certain objects over and over because of an obsession about losing them. Unlike compulsive drinking or gambling, OCD compulsions do not give the person pleasure. Rather, the rituals are performed to obtain relief from the discomfort caused by the obsessions.

3. Other features of Obsessive-Compulsive Disorder

OCD symptoms cause distress, take up time (more than an hour a day), or significantly interfere with the person's work, social life, or relationships. Most individuals with OCD recognize that their obsessions are coming from their own minds and are not just excessive worries about real problems. They realize that the compulsions they perform are excessive or unreasonable. When someone with OCD does not recognize that their beliefs and actions are unreasonable, this is called OCD with poor insight. OCD symptoms tend to wax and wane over time. Some may be little more than background noise; others may produce extremely severe distress.

Treatment

OCD can be treated in two ways: with medicine and behavior therapy. Medicines that help brain chemicals work properly can help kids with OCD. These medicines can make the obsessions and compulsions feel less intense, and they also help tone down the worry and fear. By itself, medicine doesn't fix OCD, and many kids with OCD don't need medicine to get better. But for some kids, medicine sure does help.

With or without medicine, a special type of "talk therapy" (Psychotherapy) is the most important part of treatment for kids with OCD. This type of therapy is called behavior therapy (also called cognitive-behavioral therapy). For most kids with OCD, behavior therapy helps them learn to deal with anxiety, to face fears, to resist compulsions, and to slowly but surely conquer OCD.

For more Information please visit

Obsessive Compulsive Foundation <http://www.ocfoundation.org/what-is-ocd.html>

http://www.kidshealth.org/kid/health_problems/learning_problem/ocd_p2.html

<http://www.myocd.org/>

<http://www.ocduk.org/>

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8.Phenylketonuria (PKU)

What is PKU?

PKU (phenylketonuria), is a rare, inherited metabolic disease disorder in which the body cannot metabolize the amino acid phenylalanine that is present in many

common foods. It results in mental retardation and other neurological problems when treatment is not started within the first few weeks of life. It is basically an Enzyme deficiency.

Cause

The disease arises from the absence of a single enzyme (phenylalanine hydroxylase). This enzyme normally converts the essential amino acid, phenylalanine, to another amino acid, tyrosine. Failure of the conversion to take place results in a buildup of phenylalanine. Through a mechanism that is not well understood, the excess phenylalanine is toxic to the central nervous system and causes the severe problems normally associated with PKU.

PKU is carried through an "autosomal recessive" gene. This means that two people who conceive a child must both be "silent carriers" of the gene in order for there to be a chance that the baby will have PKU.

Carrier tests are available through PKU treatment programs.

Symptoms

There may be a smaller head, and mental retardation.

Excessive build up of phenylalanine causes the skin to have a musty odor, and absence of tyrosine causes lighter skin and hair.

Diagnosis

It is done by the blood test to determine the enzyme levels.

Treatment

The treatment is only diet control for life time.

The diet for the most severe form of PKU eliminates all of the very high protein foods since all protein contains phenylalanine. Except in rare circumstances, the diet does not allow consumption of meat, fish, poultry, milk, eggs, cheese, ice cream, legumes, nuts, or many products containing regular flour. A synthetic formula is used as a nutritional substitute for the eliminated foods. The diet is supplemented with special low protein foods and weighed or measured amounts of fruits, vegetables and some grain products.

Prognosis

When a very strict diet is begun early and well-maintained, effected children can expect normal development and a normal life span. Children with the severe form of PKU once were destined to become mentally retarded and spend their lives in institutions. Children with the disease are now growing up normally. They are attending college and becoming productive adults as doctors, lawyers, teachers and engineers because of early diagnosis and strict adherence to the diet.

For more information, please visit:

[National PKU News](http://www.pkunews.org/)

<http://www.pkunews.org/>

Phenylketonuria

<http://www.ygyh.org/pku/whatisit.htm>

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9. Prader-Willi Syndrome

What is Prader-Willi syndrome (PWS)?

PWS is a complex genetic disorder that typically causes low muscle tone, short stature, incomplete sexual development, cognitive disabilities, problem behaviors, and a chronic feeling of hunger that can lead to excessive eating and life-threatening obesity.

It is named after endocrinologists Prader, Labhart, and Willi who first mentioned it.

Cause

Basically, the occurrence of PWS is due to lack of several genes on one of an individual's two chromosome 15s— the one normally contributed by the father. In the majority of cases, there is a *deletion*—the critical genes are somehow lost from the chromosome.

The critical paternal genes lacking in people with PWS have a role in the regulation of appetite. This is an area of active research in a number of laboratories around the world, since understanding this defect may be very helpful not only to those with PWS but to understanding obesity in otherwise normal people.

Symptoms

Distinctive facial features identify a child with Prader-Willi syndrome. These include a narrow face, almond-shaped eyes, small-appearing mouth, a thin upper lip with downturned corners of the mouth, and full cheeks. The child's eyes may cross .

There are associated motor problems, behavioral problems, sleep disturbances and perpetual hungry feeling. People with this flaw never feel full; they have a continuous urge to eat that they cannot learn to control.

Diagnosis

Diagnosis is made through genetic and DNA testing.

Treatment and Prognosis

There is no cure for Prader-Willi syndrome. However, the physical problems caused by the syndrome can be managed.

Treatments include food restriction, daily exercise, medication, physical and occupational therapies, speech therapy, growth hormone therapy, and special education services. If weight is controlled, life expectancy may be normal, and the individual's health and functioning can be maximized.

For more information, please visit:

Prader-Willi Syndrome Association <http://www.pwsausa.org/>

Prader-Willi Syndrome Association (UK) <http://www.pwsa-uk.demon.co.uk/>

Gene Clinic <http://www.geneclinics.org/profiles/pws/>

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10. Seizure Disorders

What is it ?

Seizure disorders are neurological disorders that may cause physical convulsions, minor physical signs, thought disturbances, or a combination of symptoms that are the result of uncontrolled electrical activity in the brain. Sometime they are symptoms of some other problem , and sometime they can appear on their own.

An individual with a seizure disorder, such as epilepsy, may experience one or more different types and levels of severity of seizure.

The condition can develop at any time of life, especially in early childhood, during adolescence and old age.

Most common word for these is Epilepsy. Epilepsy comes from a Greek word meaning "to hold or seize," and people who have epilepsy have seizures. You might also hear a seizure called a convulsion, fit, or spell.

Cause

Seizures are caused by abnormal electrical discharges in the brain. Symptoms may vary depending on the part of the brain that is involved, but seizures often cause unusual sensations, uncontrollable muscle spasms, and loss of consciousness.

Some seizures may be the result of a medical problem. Low blood sugar, infection, a head injury, accidental poisoning, or drug overdose can cause a seizure. A seizure may also be due to a brain tumor or other health problem affecting the brain. In addition, anything that results in a sudden lack of oxygen to the brain can cause a seizure. In some cases, the cause of the seizure is never discovered.

When seizures recur, it may indicate the chronic condition known as epilepsy.

Febrile seizures, relatively common in kids younger than 5 years old, can occur when a child develops a high fever, usually with the temperature rising rapidly to 102° Fahrenheit (38.9° Celsius) or more. While terrifying to parents, these seizures are usually brief and rarely cause any problems, unless the fever is associated with a serious infection, such as meningitis. A child who has a febrile seizure is not more likely to develop epilepsy

Symptoms

Most seizures occur without warning, although some people have a funny feeling, an upset stomach, or a weird smell or taste right before a seizure. This is called an aura. Others find that certain things may bring on a seizure, like not getting enough sleep or playing video games.

Even though a seizure may look scary, it's not painful. During a seizure, the person may fall down, shake, stiffen, throw up, drool, urinate (pee), or lose control of their bowels. Other seizures are less dramatic. The person may just stare into space or have jerking movements in one part of the body. When the seizure is over, the person may feel sleepy and won't remember what happened.

Diagnosis

If a person has a seizure, doctors will do some tests, such as a CAT scan, an MRI, or an electroencephalogram (EEG). A CAT scan or MRI help a doctor look at a person's brain and an EEG records brain waves. Blood tests may also be done.

All of these tests can help doctors try to find out what caused the seizure and if a person might have more seizures. But sometimes seizures are a one-time thing for a kid. Half of the kids who have one seizure never have another one.

For people who are diagnosed with epilepsy, most of them can control their seizures by taking medicines. As they get older, many kids with epilepsy get better and can stop taking medicine. For some kids, it may be difficult to get the seizures under control. A special diet or surgery may be needed.

Treatment

Treatments included seizure-preventing medicines, surgery, ketogenic diet (primarily in children), or electrical stimulation of the vagus nerve, a large nerve leading into the brain. Although seizure medications are not a cure, they control seizures in the majority of people with epilepsy.

Prognosis

For some, seizure disorders will be a temporary problem, easily controlled with medication and outgrown after a few years. For others, it may be a lifelong challenge affecting many areas of life.

It is possible to lead a normal life with this disorder. Charles Dickens the author, Vincent van Gogh the Artist, Cyclist Marion Clignet who won a silver medal at the 1996 Olympic each of them has (or had) a condition called epilepsy.

For more information, please visit:

Epilepsy Foundation <http://www.epilepsyfoundation.org/>

North Pacific Epilepsy Research <http://www.seizures.net/>

Epilepsy.com http://www.epilepsy.com/epilepsy/types_seizures.html

Neurology Channel <http://www.neurologychannel.com/seizures/>

Kid's Health http://www.kidshealth.org/parent/firstaid_safe/emergencies/seizure.html

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11. Williams Syndrome

What is William's Syndrome ?

Williams Syndrome, also known as Williams-Beuren Syndrome, is a rare genetic disorder characterized by growth delays before and after birth (prenatal and postnatal growth retardation), short stature, varying levels of mental deficiency, and distinctive facial abnormalities that typically become more pronounced with age.

Cause

Williams Syndrome is associated with deletion of genetic material in chromosome

Williams syndrome is not caused by anything the parents did or did not do either before or during pregnancy. We know that most individuals with Williams syndrome are missing genetic material on chromosome #7 including the gene that makes the protein elastin (a protein which provides strength and elasticity to vessel walls.)

In most families the child with Williams syndrome is the only one to have the condition in his or her entire extended family. However, the individual with Williams syndrome has a 50% chance of passing the disorder on to each of his or her children.

Symptoms

The disorder is characterized by the following physical features: unique elfin facial features, heart and blood vessel problems, elevated blood calcium levels, slow weight gain, feeding problems, colic, dental problems, kidney problems, hernias, and hypotonia.

Children with Williams Syndrome may be excessively social and have developmental delays, learning disabilities, and attention deficit.

Many children with WS clearly go through each day with a song (or a dance) in their hearts. Music seems to be at their fingertips, and in spite of problems in almost every other area they can be totally consumed by music of every type, and have great concentration for all things musical.

Diagnosis

The diagnosis of Williams syndrome generally has two parts:

1. Clinical diagnosis based on a variety of characteristics.
2. Medical/genetic test confirmation through a blood test: the FISH test

Treatment

Treatment typically includes physical, occupational and speech therapies.

Prognosis

Most adults with Williams Syndrome are able to complete school and hold jobs. Many live with their parents, in supervised settings or on their own.

For more information, please visit:

Williams Syndrome Association (WSA) <http://www.williams-syndrome.org/>

<http://www.williams-syndrome.org/forparents/whatiswilliams.html#1>

The Arc (a national organization on mental retardation)

1010 Wayne Ave

Suite 650

Silver Spring, MD 20910

Tel: (301)565-3842

Fax: (301)565-3843

Tel: (800)433-5255

Email: info@thearc.org

Internet: <http://www.thearc.org/>

Williams Syndrome Foundation

161 High Street

Tonbridge, TN9 1BX

United Kingdom

Tel: 01732 365152

Fax: 01732 360178

Email: John.nelson-wsfoundation@btinternet.com

Internet: <http://www.williams-syndrome.org.uk>

Canadian Association for Williams Syndrome

Tel: 6048530231

Fax: 6048530232

Email: sev@uniserve.com Internet: <http://www.caws-can.org>

NIH/National Institute of Child Health and Human Development

Internet: <http://www.nih.gov/hichd/>

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12.Disorders caused by Injuries to the Brain

12.i Cerebral Palsy(CP)

What is CP?

Cerebral palsy (CP) is a disorder that affects muscle tone, movement, and motor skills (the ability to move in a coordinated and purposeful way). Cerebral palsy can also lead to other health issues, including vision, hearing, and speech problems, and learning disabilities.

CP is usually caused by brain damage that occurs before or during a child's birth, or during the first 3 to 5 years of a child's life. There is no cure for CP, but treatment, therapy, special equipment, and, in some cases, surgery can help a child who is living with the condition

Cerebral means having to do with the brain. **Palsy** means weakness or problems with using the muscles.

Causes

These disorders are caused by damage to a child's brain early in the course of development. The damage can occur during fetal development, during the birth process or during the first few months after birth. Although cerebral palsy affects movement, the underlying problem originates in the brain, not in the muscles themselves

Low birth weight and premature deliveries are the also prime risk factors.

Three other factors that contribute to the risk of CP, regardless of birth weight, include congenital malformations, inflammation of the placenta, and twinning.

A small number of children with cerebral palsy acquire the disorder after birth. In these cases, doctors can sometimes pinpoint a specific reason for the neurological problem. For example, cerebral palsy can develop after an illness during early infancy, such as bacterial meningitis — an infection and inflammation of the membranes and fluid surrounding the brain and spinal cord. It can also be the result of a head injury.

However, doctors don't completely understand the cause of most cases of cerebral palsy, which are present at birth (congenital). For many years, doctors and researchers believed that cerebral palsy was caused by a lack of oxygen during birth. Now they believe that only a small number of cases are caused by problems during labor and delivery.

Some possible causes

Researchers have now identified many possible causes of congenital cerebral palsy, including:

- **Maternal infection during pregnancy**, such as rubella or other viral infections
- **Severe jaundice in newborns**, which may be caused by infection, severe bruising or problems with red blood cells due to ABO or Rh incompatibility — two incompatibilities between the blood of the mother and her fetus
- **Abnormal brain development before birth**, resulting from genetic causes or metabolic disorders
- **Disturbance to brain circulation before birth**, caused by an artery spasm or blood clot, similar to a stroke in adults

Symptoms

It can be mild, moderate, or severe.

Mild CP may mean a child is clumsy.

Moderate CP may mean the child walks with a limp. He or she may need a special leg brace or a cane.

More severe CP can affect all parts of a child's physical abilities. A child with moderate or severe CP may have to use a wheelchair and other special equipment.

Sometimes children with CP can also have learning problems, problems with hearing or seeing (called *sensory problems*), epileptic seizures, or mental retardation. Usually, the greater the injury to the brain, the more severe the CP. However, CP doesn't get worse over time, and most children with CP have a normal life span.

Types of CP

There are three main types of CP:

- **Spastic CP** is where there is too much muscle tone or tightness. Movements are stiff, especially in the legs, arms, and/or back. Children with this form of CP move their legs awkwardly, turning in or scissoring their legs as they try to walk. This is the most common form of CP.

- **Athetoid CP** (also called **dyskinetic CP**) can affect movements of the entire body. Typically, this form of CP involves slow, uncontrolled body movements and low muscle tone that makes it hard for the person to sit straight and walk.
- **Mixed CP** is a combination of the symptoms listed above. A child with mixed CP has both high and low tone muscle. Some muscles are too tight, and others are too loose, creating a mix of stiffness and involuntary movements.

More words used to describe the different types of CP include:

- **Diplegia**--This means only the legs are affected.
- **Hemiplegia**--This means one half of the body (such as the right arm and leg) is affected.
- **Quadriplegia**--This means both arms and legs are affected, sometimes including the facial muscles and torso

Diagnosis

Early signs of cerebral palsy may be present from birth, but it's often difficult to make a definite diagnosis during the first six months. Cerebral palsy is generally diagnosed by age 1 or 2.

Many of the normal developmental milestones, such as reaching for toys (3-4 months), sitting (6-7 months), and walking (10-14 months), are based on motor function. A physician may suspect cerebral palsy in a child whose development of these skills is delayed

If your child shows some signs of cerebral palsy, your doctor will likely schedule an appointment to observe your child and to talk to you about your child's physical and behavioral development. In this evaluation, your doctor will check your child's reflexes, muscle tone and movements. Additional tests may rule out other disorders that can cause movement problems. Your doctor may have your child undergo one or more of these procedures:

- **Computerized tomography (CT) scan.** Images created with a CT scanner show the structure of your child's brain, as well as the presence and extent of any damage.
- **Magnetic resonance imaging (MRI) scan.** The cylinder-shaped MRI scanner uses no X-rays. Instead, a computer creates tissue-slice images of the brain from data generated by a powerful magnetic field and radio waves. These images can be viewed from any direction or plane.
- **Other tests.** Some children may need genetic or metabolic tests to help rule out other conditions.

You may be referred to a specialist to help determine if your child has cerebral palsy or some other condition. Your doctor may recommend a visit to a doctor with specialized training in the development of the brain and nervous system in children (pediatric neurologist), a doctor who specializes in childhood development (developmental pediatrician), or a doctor who specializes in physical medicine and rehabilitation (physiatrist)

Treatment

Currently there's no cure for cerebral palsy, but if your child is diagnosed with it, a variety of resources and therapies can provide help and improve the quality of your child's life. Many children learn how to get their bodies to work for them in other ways. For example, one infant whose CP keeps him from crawling may be able to get around by rolling from place to place.

With early and ongoing treatment the effects of CP can be reduced. Typically, children with CP may need different kinds of therapy, including:

- **Physical therapy** (PT), which helps the child develop stronger muscles such as those in the legs and trunk. Through PT, the child works on skills such as walking, sitting, and keeping his or her balance.
- **Occupational therapy** (OT), which helps the child develop fine motor skills such as dressing, feeding, writing, and other daily living tasks.
- **Speech-language pathology** (S/L), which helps the child develop his or her communication skills. The child may work in particular on speaking, which may be difficult due to problems with muscle tone of the tongue and throat.

The child may also find a variety of special equipment helpful. For example, braces may be used to hold the foot in place when the child stands or walks. Custom splints can provide support to help a child use his or her hands. A variety of therapy equipment and adapted toys are available to help children play and have fun while they are working their bodies. Activities such as swimming or horseback riding can help strengthen weaker muscles and relax the tighter ones.

In addition, medication and surgery can help improve muscle function. Surgery can help repair dislocated hips and scoliosis (curvature of the spine), which are common problems associated with CP. Severe muscle spasticity can sometimes be helped with medication taken by mouth or administered via a pump (the baclofen pump) implanted under the skin.

Prognosis

Most people with cerebral palsy experience a normal life span. Those with severe forms of CP may have a reduced life span. As people with cerebral palsy age,

they may experience long-term effects of chronic physical impairment, such as the following:

- Increase in spasms
- Increase in shortening of muscles (contractures)
- Joint problems (e.g., pain, loss of flexibility)
- Tight muscles
- Increase in back pain
- Emergence of incontinence
- Increase in incontinence
- Reduced energy levels

Ways to maintain physical function include the following:

- Appropriate wheelchair seating and posture
- Assume various positions out of the wheelchair
- Use a wheelchair when fatigued and when walking is difficult
- Regular and appropriate exercise, including stretching exercises and exercises to maintain flexibility in joints
- Maintain ideal weight
- Have regular medical check-ups
- Avoid sustained mental stress
- Carefully consider proposed surgery
- Plan for appropriate rehabilitation after surgery

For more information, please visit:

<http://www.ucpcleveland.org/home1.asp>

National Institute of Neurological Disorders and Stroke at the National Institutes of Health www.ninds.nih.gov

www.dreamms.org

a nonprofit information clearinghouse on assistive technology

www.lburkhart.com

ideas and instructions for adapting toys for use by children with CP

Medical university of South Carolina

<http://www.mushealth.com/gs/healthtopic.aspx?action=showpage&pageid=P02561>

Mayo clinic

<http://www.mayoclinic.com/health/cerebral-palsy/DS00302/DSECTION=6>

Kid's Health

http://www.kidshealth.org/parent/medical/brain/cerebral_palsy.html

United Cerebral Palsy Associations, Inc.

1660 L Street NW, Suite 700

Washington DC 20036

800.872.5827 *Voice/TTY*

202.776.0406 *Voice*

www.ucp.org *Web*

Easter Seals--National Office

230 West Monroe Street, Suite 1800

Chicago, IL 60606-4802

800.221.6827 *Voice*

312.726.6200 *Voice*

www.easter-seals.org

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12.ii. Neural Tube Defects (Spina Bifida)

What is Spina Bifida

Spina Bifida is a permanently disabling birth defect. It is a Neural Tube Defect, and is one of the most devastating of all birth defects

Spina Bifida or Neural tube defects are birth defects that involve the central nervous system. These defects of the spinal cord and/or brain result from failure of the neural tube to properly form.

Cause

It results from the failure of the spine to close properly during the first month of pregnancy. In severe cases, the spinal cord protrudes through the back and may be covered by skin or a thin membrane.

Diagnosis

It may involve loss of sensation and severe muscle weakness of the body below the level of the lesion. Although outcomes vary widely, with new medical treatments and technology, many people with spina bifida can expect to live a normal life, and often have careers, get married, and have families.

These disorders may present varying degrees of disability, including learning disabilities, social issues, lower extremity paralysis, loss of bowel or bladder control, and hydrocephalus (water on the brain), which can produce retardation unless it is surgically treated.

Treatment and Prognosis

Surgery to close a newborn's back is generally performed within 24 hours after birth to minimize the risk of infection and to preserve existing function in the spinal cord.

Most children born with Spina Bifida live well into adulthood as a result of today's sophisticated medical techniques.

For more information, please visit:

[Spina Bifida Association of America](#)

http://www.sbaa.org/site/c.gpILKXOEJqG/b.2016945/k.2321/Spina_Bifida_Association_Web_site.htm

Indian group –

Mrs. Vinita Jindel
G-14, Krishna Marg, C-Scheme
Jaipur-302001 (Raj) India.
Or Mail Us At: c_jindel@datainfosys.net

<http://www.indiaspinabifidaassociation.org/>

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12.iii. Traumatic Brain Injury (TBI)

Introduction

Traumatic Brain Injury (TBI) is a disability category that occurs as the result of an injury to the brain as a result of an accident, insufficient oxygen, poisoning or infection at any time during an individual's life. It does not include congenital or degenerative brain injuries or brain injuries caused by birth trauma.

The severity of brain injuries can vary greatly, depending on the part of the brain affected and the extent of the damage. A mild brain injury may cause temporary confusion and headache, but a serious one can be fatal.

Traumatic brain injuries may result in impairment in cognition, language, social skills, memory, attention, reasoning, behavior, physical functioning, psychological functioning, information processing, or speech. Physical challenges can include ambulation, balance, coordination, fine motor skills, strength, and endurance. Medical and neurology specialists determine a diagnosis using behavioral and neuropsychological assessments. In addition to rehabilitation services, individuals with TBI are treated with many of the services and supports as individuals with developmental disabilities. Outcomes vary by level of injury, with the goal for many being to return to school or to work, but with new and different levels of support.

Causes

Brain injury can occur in many ways. Traumatic brain injuries typically result from accidents in which the head strikes an object. This is the most common type of traumatic brain injury. However, other brain injuries, such as those caused by insufficient oxygen, poisoning, or infection, can cause similar deficits.

Symptoms

Your brain controls your movements, behaviors, thoughts and sensations. Not surprisingly, then, a brain injury can affect many different aspects of your physical and emotional well-being.

Most symptoms of a brain injury appear immediately or shortly after a blow to the head. In many cases, however, the symptoms are subtle and easy to miss — even for the person who's been injured.

Signs and symptoms of a mild brain injury can include:

- Brief period of unconsciousness
- Headache
- Confusion
- Dizziness
- Sensory problems, such as blurred vision, ringing in the ears or a bad taste in the mouth
- Mood changes
- Memory or concentration problems

If the injury is moderate to severe, the list of signs and symptoms grows to include:

- Persistent headache
- Repeated vomiting or nausea
- Convulsions or seizures
- Inability to awaken from sleep

- Dilation of one or both pupils of the eyes
- Slurred speech
- Weakness or numbness in the extremities
- Loss of coordination
- Increased confusion or agitation

Children with brain injuries may lack the communication skills to report headaches, sensory problems, confusion and similar symptoms. Instead, they may refuse to eat and appear listless or cranky. Their sleep patterns and school performance may change, and they may lose interest in favorite toys or activities.

Problems associated with traumatic brain injuries often come in two stages. The original impact may bruise portions of the brain or directly sever nerve connections. The second stage of the injury occurs when the tissue at the injury site begins to swell.

It is normal for injuries to cause swelling. Think of the last time you got a bad bruise. In addition to being colorful, the bruised area probably became tender and swollen. Now imagine that happening on a larger scale inside a bony confined space.

Inside the skull, there's no place for this swelling to go, so the pressure on the brain increases. Intracranial pressure must be monitored closely because it can result in additional damage to the brain.

Diagnosis

Emergency medical personnel assess the severity of a brain injury by seeing how well the injured person can follow directions to blink his or her eyes or to move extremities. The coherence of the person's speech also provides important clues.

Imaging tests often are crucial in determining the extent of injury. Skull and neck X-rays can check for bone fractures or spinal instability. Computerized tomography (CT) scans can uncover evidence of:

- Bleeding in the brain (hemorrhage)
- Large blood clots (hematomas)
- Bruised brain tissue (contusions)
- Brain tissue swelling

Treatment

Fewer than half the people who suffer severe traumatic brain injuries need surgery to remove or repair the damaged portions of their brains. In some cases, there is a collection of blood between the skull and the brain. This is called an intracranial hematoma, which must be surgically drained.

Surgery may also be performed to drain the excess fluid that has accumulated in reaction to the trauma itself. While swelling is a natural reaction for body tissue that has been injured, it can cause additional damage to the brain by increasing the pressure inside the skull. Medications also can be used to decrease this pressure.

Most people who have had a significant brain injury will require rehabilitation. They may need to relearn basic skills, such as walking or talking. The overall goal is to improve their abilities to function at home and in the community.

Prognosis

A brain injury often erases your memory of events that occurred just before your injury. It can also make it more difficult for you to remember new information and learn new tasks. While some memory problems may diminish over time, others may be permanent.

As they slowly recover and adjust, some people who have had brain injuries carry cue cards that list coping strategies, such as:

- Slow down.
- Stop and think.
- Break it down, step by step.
- Ask questions.
- Do not assume.
- Pay attention to details.
- Take a break.
- Think of other things to try to problem solve.

Many people also find it helpful to carry a calendar for planning and organizing daily activities, step by step. It can serve as a reminder of uncompleted tasks and as a storage site for information you might otherwise forget.

For more information, please visit:

Traumatic Brain Injury Resource Guide <http://www.neuroskills.com/>

Mayo Clinic <http://www.mayoclinic.com/health/traumatic-brain-injury/DS00552/DSECTION=3>

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