BIRTH DEFECTS AND DEVELOPMENTAL DISABILITIES

Objectives:

At the end of this class, you should be able to:

1. Describe the causes, signs and care of the person with autism, Down syndrome, cerebral palsy, hearing loss, vision loss, mental retardation, and spina bifida.

Introduction

Birth defects and a large number of developmental disabilities happen before the baby is born. Most occur during the first 3 months of the mother's pregnancy. They affect how the body looks, how the body works and both looks and body function. Some are easy to see immediately after birth, such as a cleft lip and a club foot. Others, such as a heart defect or cerebral palsy, may not be seen until some time after a baby is born.

They may be seen only after the young baby grows or with special tests. Some are very mild and others may be very severe. Some new babies die from them. Many lower the person's ability to talk, hear, learn, help themselves and live on their own without help. Surgery and treatments are often needed. There is no cure. They last the person's entire life.

One of every 33 babies is born with a birth defect. The most common is a heart problem. About 1 in every 100 to 200 babies is born with a heart problem.

Some examples are:

- autism;
- Down syndrome;
- cerebral palsy;
- hearing loss;
- vision problems;
- mental retardation;
- spina bifida and
• seizures (epilepsy).

What Causes It?

Some women have more of a chance of having a child with a birth defect than others. These are some of the things that make some people more at risk than others. These things are:

• having a baby after the mother is over 35 years old;
• the use of illegal and some prescription drugs;
• smoking and drinking alcohol during pregnancy;
• a family history of birth defects.

Some of the things that can be done to prevent some birth defects and developmental disabilities include:

• genetic counseling before getting pregnant;
• no drinking or smoking;
• not taking any medicines or illegal drugs;
• good medical care during pregnancy and
• taking vitamin B.

Autism

Autism is a developmental disability that:

1. lowers the person's ability to communicate with others;
2. leads to poor social interactions with others;
3. causes the person to have unusual repetitive behaviors, like rocking, and narrow interests.

Some may have mild signs. Others may be severe. These people may have very different ways of learning, paying attention and reacting to things. Mental ability may be very high or very low. It begins before 3 years of age, but, it is sometimes seen
when the child is only 1½ years old. It lasts for the person’s entire life. It can occur in anyone, but boys are four times more likely than girls to have it. It was found that 1 in 150 U.S. children who are 8 years old have autism. It runs in some families. Many families may have more than one child with it. Early treatment helps these children very much.

The Signs

Children with autism may:

- not respond to their name;
- not make eye contact with others;
- not understand what others are feeling or thinking;
- start to talk later than others without it;
- not refer to themselves as "I" but instead by name, like Jane or Jim;
- do self harm with things like head banging;
- not want to play with others;
- sing-song about their favorite thing;
- not be able to feel pain like other children do;
- react differently to sounds and touch;
- not want to be hugged;
- also have other medical problems like a brain tumor, epilepsy, mental retardation and/or an attention deficit disorder.

What Causes It?

We are not sure what causes it. It may come from a number of things, such as:

- genes;
- environment; and
- childhood vaccines.

Some say that many things may cause it. It seems to run in families and it is seen more when the child has:

- fragile X syndrome;
- mental retardation;
- seizures;
- sleep problems;
- poor muscles;
- a lack of feeling pain;
- a lack of feeling cold;
- measles (rubella) while in the mother's body.

It is also seen more often when the mother has:

- taken some drugs during her pregnancy;
- been exposed to mercury or bug killers;
- not had enough vitamins, like zinc.
There is no way to prevent it because the cause of it is not clear. There is also no cure for it, but these children can be helped with special things.

_Caring for the Person with Autism_

One of the biggest problems is talking with others. These children do not learn to talk as soon as others. Some of the things that you can do with these children to help them to talk are:

- using signs and pointing to objects to communicate;
- using pictures or a picture board;
- singing along with songs on a video or CD; and
- swinging on a swing while learning words.

It is also treated with:

- keeping the child free of dangers and cold. They may not feel pain or cold;
- routines and care that you stick to;
- vitamins and minerals like vitamins B6 and C;
- special diets that do not have wheat or dairy foods;
- food allergy testing and treatment;
- drugs to treat the symptoms like fighting, self-injury, anxiety, depression, and attention problems;
- speech therapy;
- physical therapy;
- listening help;
- self-help teaching;
- music and art;
- animal therapy, like riding a horse;
- occupational therapy;
- behavior therapy; and
- clearing the body of heavy metals.

Many of these children are able to grow up and live a nice life with help. Some may be able to live on their own, with help. Others may do better in a group home.

_Down Syndrome_
Down syndrome has been around for a long time. A bad 21st chromosome is the cause. It can be mild or very severe.

It is found in about 1 out of 800 to 1,000 births in the U.S. About 350,000 people in our nation have this gene birth defect. People of all ages and races may have it. Many are able to live almost normal lives with treatment, education and the support of others, like their family and others in the area where they live. Some are able to go to high school and college. Some can find jobs and live on their own. All of these people should be able to do the best that they can do with the help of other people.

The Signs

Down syndrome is known before or just after the new baby is born. The face is most often noted after birth. The face can be flat, the ears may be small, the tongue may be large and the eyes may have a slant up.

Some of the other signs of this birth defect are:

- mental retardation;
- a different look of the person's face;
- weak "floppy" muscles;
- weak joints;
- small hands and feet;
- heart problems;
- hearing loss;
- poor speech;
- slow acting thyroid gland in the neck; and
- small white spots of the eye;

What Causes It?

Babies normally get a total of 46 chromosomes. They get 23 from the mother and 23 from the father. When the baby gets 47, and not 46, the baby is affected with physical and mental problems.

It is known that mothers, after the age of 35, are more likely to have a baby with this Downs. There is 1 chance in 1,000 when the mother is 30 years old. By age 35, the chances increase to 1 in 400. At age 42, the chances rise again to 1 in about 60 babies.

Caring for the Person with Down Syndrome

Children often have speech, mental and physical problems. They may grow at a slower rate. They may have problems with talking, thinking and learning. They may also have eating, digestion, heart, hearing, weight and sight problems.

Children can do well but they do need help. They often need a lot of medical care for their physical problems, such as heart disease, eye problems, and digestion problems. They may also do well with speech, physical and occupational therapy. They often attend special classes in their school or a special school that meets their needs.
Care givers and parents must:

- know the child, their likes and dislikes;
- provide medical care, as the doctor orders;
- encourage the child to do as much as they can for themselves;
- give the child a chance to do things for themselves, like dressing, brushing teeth and other activities of daily living;
- give praise when the child does well;
- support their education; and
- follow the orders of the physical, occupational, and speech therapists.

Cerebral Palsy

Cerebral palsy is often called simply CP. Children with CP have trouble when they try to walk, crawl or grab onto things. Some children may also have trouble with vision, speech, and hearing. Others may have seizures, poor mental ability, trouble with bowel and bladder control, eating and feeding. CP is a disorder. It is not a disease.

The term CP includes several muscle problems all of which result from damage to one or more parts of the person's brain. This damage can happen before birth or up to the age of 5 years old. There is no cure for CP but good care can help the person to function in the best way possible for the person.

CP is rare. Less than 2 out of 1,000 children have CP. About one in 100 babies who are small or premature have CP. In the United States, 764,000 people have one or more signs of CP. 8,000 babies and 1,200 to 1,500 pre-school children are diagnosed with CP every year.

The Signs

Children with CP are not able to control their muscles and their coordination. Muscles get tight. The person loses voluntary movement. They may have walking problems, swallowing and speech problems depending on what part of the brain has been damaged.

There are 4 kinds of CP. They are:

1. Spastic,
2. Athetoid,
3. Ataxic, and
4. Mixed forms.

Spastic CP
About 70% to 80% of CP cases are spastic CP. This kind of CP can be mild or it can be very severe. The arms and/or legs are not fully developed. The muscles are weak, stiff and tense. At times the arms or legs become so tight and tense that they bend up against the body (contracture). In the beginning, muscles are weak and without tone. After that, the muscles become stiff and tense.

Some children who have the mild form of this kind of CP may have problems during only some activities. Some may only have a problem when they run or walk. Those with a severe form of this CP can have paralysis of:

- both legs (diplegia),
- one side of the body (hemi-paresis) or
- the entire body.

Toe walking and a scissor walk are seen in this kind of CP. Speech problems are also seen. One kind of speech problem called dysarthria is sometimes present. These children are not able to say words in the correct way.

Athetoid CP

This kind of CP is seen in 20% of children with CP. Slow, unusual, and involuntary movement is seen in the hands, arms, legs and feet as well as with the body's trunk. Jerky and sudden movements may occur. The face and the tongue can also be affected. The person may make strange faces (grimace) and drool saliva from their mouth. They can also have a problem with talking. The person may not be able to speak words in the correct way.

These movements happen mostly when the person is nervous and tense. They go away when the person is sleeping or resting.

Ataxic CP

This kind of CP is seen in only about 5% to 10% of children with CP. It is rare. It affects the person's balance. The child may only be able to walk with their feet far apart so that they do not fall. They may also have trouble doing things like reaching for a toy or writing their name. The child's hand may begin to shake very fast when they try to touch a toy or try to write their name.

This kind of CP may lead to:

- weak muscles,
- an unsteady walk,
- rapid movements, and
- no control over this movement.

Mixed Form CP

A few children may have more than one form of CP. For example, a child may have both athetoid and spastic CP.
Doctor's and parents do not always know that the new baby has CP until months after they are born. In fact, the signs of CP may not be seen until the child is a year old. The signs, however, do appear by the time the child is 1½ years old.

CP is first seen when the baby or young child is not able to crawl, walk, sit, and roll over at the age they are expected to. CP signs are seen when an 8-month-old baby is still not able to roll over. It may be first seen when an 18-month child is still not able to crawl on the floor.

Other early signs are:

- weakness on one side of the body
- a sitting position that is not normal
- muscles that do not have a normal tone or firmness (hypotonia)
- muscles that are very stiff (hypertonia)

Muscles are floppy and weak when they do have little tone. Muscles are stiff and rigid when they are hypertonic. Babies with CP may start their life with floppy muscles. Their muscles may then become stiff after a couple of months of life.

What Causes It?

The cause of it is still not very clear. But, CP is found in infants and children that:

- are born before the full 9 months of pregnancy;
- are born very small and with a low birth weight;
- have a blood type or a blood factor that is different from their mother's;
- have a mother who got an infection like the German measles when they were pregnant, especially during the early stage;
- had a physical or growth problem before they were born;
- had trouble and trauma during birth;
- got a yellow color of the skin and liver problems (jaundice) soon after birth;
- have brain damage just before birth, during birth or just after birth; or
- had a lack of oxygen during birth, just before birth or after birth.

About 70% of CP happens because the child's brain was damaged in the mother's uterus before they were born. Why this has happened is not clearly known.

CP can also come from a bad injury after birth if the young child:

- does not get enough fluid and becomes dehydrated, or
- gets a bad infection of the brain or spinal cord, or
- had a bad car accident, or
- had a bad fall.

Some cases of CP can be prevented when women who can have babies get the proper treatment and care.

Some of the things are:

- get the measles shot;
• get the Rh factor shot 72 hours after a baby is born so that her next babies will not get CP;
• get good care so that the baby will not be born too early;
• stay away from infections;
• take only medicines that are necessary;
• avoid X-rays;
• get good nutrition;
• get treatment for diabetes and anemia if they have it and
• keep the baby safe so they do not have an accident, like a fall.

Caring for the Person With CP

CP patients can lead close to normal lives with treatment and care. The goal of care for these patients is to help them be as independent as they can. Care should be started right after the person has been diagnosed with CP.

The health care team that works with these children includes:

• doctors,
• nurses,
• physical therapists,
• occupational therapists,
• therapy aides,
• speech therapists,
• social workers
• eye doctors,
• personal care aides,
• teachers, and
• family members.

Children with mild mental and physical problems can, and should, go to regular schools. Others may have to go to a special school. Total independence is often not possible for some. They may need help with certain things, like the activities of daily living, for their whole life.

All patients with CP should do as much as they can. This helps to increase the person’s independence and self esteem. It also lowers the work load for the family members.

Some patients with CP may also get:

• medicine,
• surgery to improve muscles and prevent further lack of function;
• braces for support and to prevent further loss of function; and/or
• restorative/rehabilitation care.

A doctor writes an order for restorative/rehabilitation care. After this, the physical therapist, occupational therapist and/or speech therapist see the patient or resident so that they can:
• think about and find the best way to go about the care that the doctor has ordered and
• decide on the goals of the care for the patient or resident.

The goals can include being able to:

• eat on their own using special plates and utensils if needed,
• get in and out of the bath tub or shower with a shower chair and grab rails if needed,
• climb up stairs safely,
• grab things,
• have increased muscle strength, and
• communicate with others with the spoken word.

Patients work with the healthcare team to decide on these goals. The person with CP must be work to reach these goals on a daily basis. How well the person is reaching these goals is documented on a flow sheet, progress note or another tool, as used at your place of work.

Most of this care is provided by physical therapists, occupational therapists and speech therapists. Some rehab centers also have assistants and aides to help with this care. These people are given special training so that they are able to help the person to reach their goals.

Some of the devices that are used in this care are:

• splints,
• walkers,
• canes,
• shower chairs,
• grab bars on the side and the back of the bathtub or toilet,
• graspers or reachers to lift items up from the floor,
• special eating utensils with built-up handles to help the person feed themselves,
• special combs and brushes so that the person can groom themself,
• shoehorns to help a person get dressed even though they do not have full range of motion,
• raised sitting chairs, raised toilet seats, and chair leg extenders help people safely transfer without the help of another person and
• cups with lids and special plates with deep centers and weight help people eat their meal without spills.

Rehabilitation and restorative care play a very important part of healthcare.

A chronic disability, like CP, affects about 10% of all children in the United States. Many of these children have pain. They have to go to the doctor's office often and they go to the hospital more than children who do not have a disability. It is hard for them to play and interact with other children. Other children may avoid them and say unkind things to them.
Parents of a child with a disability may feel guilty about having a less than perfect child. These parents may get angry, depressed, sad and guilty. They may be over worked with the care that the child needs.

These parents also need the help of healthcare professionals. They need to be taught about the child's health problem and how to care for them. One source of help and information for parents is the United Cerebral Palsy Association.

**Hearing Loss**

Hearing loss can be very mild, mild, moderate, severe or very severe. The term "deaf" is used for someone that has a severe hearing loss and "hard of hearing" is describe people who have a less severe hearing loss than deafness.

About 72,000 children, from 6 to 21 years old, have a hearing impairment" according to their school. Another 1,600 children got services under the "deaf blind" category. 9 in every 10,000 children ages 3 to 10 years had a moderate to high hearing loss in both ears.

Hearing loss is more common among older children than among younger children. Sadly, 30% of children with a hearing loss have one or more other problems, like CP and mental retardation, in addition to their hearing loss. Boys have more hearing loss than girls.

**The Signs**

Hearing loss can be mild or it can be very severe. Hearing loss is sometimes found right after the baby is born. At other times, it is found later in life as the child grows. This loss is found in new babies when they do not react to loud noises with a natural startle reflex. It is found later in life by the doctor or by the child's school.

Children with this kind of loss may not be able to talk like others and they may not be able to communicate with others like other children do. They may also have learning problems in school because they cannot hear the teacher.

**What Causes It?**

Normal hearing happens when all the body parts for hearing are in good shape. The parts of the body that are needed for good hearing are:

- the outer ear,
- the middle part of the ear,
- the inner part of the ear,
- hearing nerves and their ability to work with the brain.

Hearing loss may occur before the baby is born as a result of a bad # 26 gene and from infections, like rubella (measles). They can also happen during the child's birth if the baby:

- does not get enough oxygen,
- has bleeding in the brain, and/or
- has a severe case of jaundice.

Hearing loss seems to happen more often when the baby is born too soon and when they have a low birth weight.

This loss can also happen as the child is growing up. Some of these causes are:

- infections like ear infections and measles,
- head injuries,
- some antibiotics, and
- constant loud noises.

Some causes of hearing loss can be stopped with things like a vaccine to prevent measles in new mothers and light therapy to treat any jaundice after the new baby is born.

**Caring for the Person With a Hearing Loss**

The care of this person may include the below, as based on the person's own needs and the amount of loss:

- speak clearly and slowly while facing the person. The person may be able to read your lips as you speak;
- use pictures to communicate;
- use signals and sign language if you and the child know it;
- give the person their hearing aid, if they have one; and
- help the child to learn and speak, using things that work well for the child.

**Vision Loss**

![Glasses](image)

Vision loss can lead to a person's not seeing things clearly or not seeing anything at all. Some may also not see as wide an area as they should unless they turn their head or move their eyes to the side.

Vision loss is seen more with old adults than in children. Many adults get bad vision as they get older. Diabetes can lead to vision loss and even blindness in adults. Only 1.4 out of 1,000 8 year olds have low vision or blindness. Many of these children also have other birth defects and disabilities like CP and mental retardation.

**The Signs**
Severe vision problems are sometimes found soon after the person is born. The baby may not be able to follow a moving object, like a toy, or the mother's face or finger. At times, nobody knows the child has a vision problem until they go to school and they are having problems with learning.

What Causes It?

Vision loss can happen before birth and at any time throughout the person's life. It happens when:

- there is damage to the eye;
- the shape of the eye is not normal;
- the brain does not work right when it has to process what the person sees.

A baby that is born too soon and those that have a low weight when they are born are more likely to have a vision loss than other babies.

Other common causes are:

- a bad gene,
- a lack of oxygen before and during birth,
- too much fluid in the brain,
- infections, and
- albinism (pale, white skin color and red eyes)

Caring for the Person With a Vision Loss

The care of this person may include the below, as based on the person's own needs and the amount of loss:

- make sure that the person is safe;
- keep the room neat and orderly so the person does not trip and fall;
- give the person their eye glasses, if they have one;
- help the child to read, using things like Braille raised symbols.

Mental Retardation

About 1 in 100 children from age 3 to 10 years have this disorder. It is found more in boys than in girls. It is also found more in black children than in white children. Mild mental retardation is more common than a severe degree of it. In the U.S. it is found most often in West Virginia and the least often found in Alaska.

The Signs

Children with this birth defect have low mental ability and some poor self-care skills. They have a hard time learning new things. It may take them a longer time to learn a new thing than it does with other children. They may also not be able to get dressed and relate to others. It can be very mild or it can be very severe.

What Causes It?
For most, the cause is not known. But, at times, it can be caused by:

- a disease,
- an injury,
- fragile X syndrome,
- bad genes,
- infections,
- Down syndrome;
- extra fluid on the brain;
- low oxygen to the baby during birth,
- the mother drinking alcohol while she is carrying the baby;
- a head injury,
- PKU (phenylketonuria) at birth, which can be treated,
- jaundice at birth, which can be treated,
- stroke, or
- another problem with the brain.

*Caring for the Person With Mental Retardation*

Good care of the child includes:

- keeping the child safe,
- teach things in a slow manner,
- repeat things when needed,
- assist the person with their daily routines, like dressing and bathing,
- praise and rewards when the person does well.

These children often need the help of an occupational therapist and special teachers. Many are able to hold simple jobs and live alone in their own home or in a special group home.

*Spina Bifida*

Spina Bifida is seen in about 7 out of every 10,000 births in the U.S. Some mothers end the pregnancy before the baby is born if they find out that the baby has it.

*The Signs*
There are 3 kinds of this birth defect. They are:

1. Hidden. There is no hole or opening on the back. The spinal cord and nerves are usually normal but there are small spaces between a few of spine bones. Later in life, the person may have minor nerve problems.
2. Meningocele. This form is usually mild with no nerve damage or only minor problems later in life. There is a sac that is pushed through the open part of the spine.
3. Myelomeningocele. This is a severe form. The covering of the spinal cord and the nerves push out from the back. Severe nerve damage and other problems occur.

It is found more with white women than black women and more with Hispanic women than non-Hispanic women. It is also found more with families that are poor.

It can lead to:

- learning problems, as a result of excess fluid on the brain;
- urinary problems (incontinence);
- bowel problems (incontinence) and
- social problems.

What Causes It?

It can be caused by a number of things, such as the below during pregnancy:

- problems when the nervous system is developing;
- lack of vitamins, including vitamin B;
- insulin dependent diabetes;
- the use of some medications for seizures;
- obesity;
- fevers;

Taking vitamin B (folic acid) every day before and during pregnancy can prevent it for some people.

Caring for the Person With Spina Bifida

Good care of the child includes:

- keeping the child safe,
- teaching things in a slow manner,
- repeating things when needed,
- assisting the person with their daily routines, like dressing and bathing,
- bowel and bladder programs,
- physical care of urinary and bowel needs (catheters, enemas, etc.)
- praise and rewards when the person does well.

These children often need the help of an occupational therapist and special teachers. Many are able to hold simple jobs and live alone in their own home or in a special group home.
Seizures

Seizures happen when the brain does not work in the correct way. The person may have unusual movements and changes in their level of awareness and consciousness. Some may only affect one part of the body. Others can affect the whole body.

About 3 in 100 children will have one seizure before they reach the age of 15. High fevers lead to about ½ of these cases. 1 in every 100 children has seizures that occur often. This is called epilepsy. Many have this for their entire life, but others outgrow it.

The Signs

A new baby may not have convulsions. Their eyes may simply be looking in different directions and their lips may smack. They may also stop breathing.

Older children with may have a simple, partial seizure that occurs in one part of the body and then moves to other parts of the body. They remain awake. Other people may have a complex, partial seizure. The person repeats a thing like clapping and they cannot remember the event after it is over. Still others may have a general convulsive or general nonconvulsive type.

There is muscle jerking for several minutes that cannot be controlled during a convulsion. The person may incontinent of urine and stool and they will be tired after it. At times, the person can get hurt from the jerking with such things as a broken bone or a bite on the tongue. When a seizure continues for more than 15 minutes it is very dangerous. 911 should be called to treat the person.

What Causes It?

The cause for most is not known. It runs in families and it may be caused by one or more of the below:

- infections;
- developmental problems like CP;
- head injuries;
- problems with metabolism;
- drugs;
- some medicines;
- poisons;
- bleeding in the brain; and
- poor blood supply to the brain

Caring for the Person With Seizures

Many people do well with special medicines to prevent seizures. It is important that the person get their medicine in the correct dose as the doctor has ordered.

You must protect the person by doing the below during a seizure:
• Help the child to lie down so they do not fall.
• Take off their eye glasses.
• Remove any hard or dangerous items from the area.
• Do NOT put anything at all in the person's mouth.
• Check for breathing. Call for help with 911 and immediately begin rescue breathing or CPR, when needed, as long as the convulsion allows you to do it without injury to the person or yourself.
• Lie the person on their side and watch them until they are fully awake after it.

Summary

It is sad to know that so many children are born with a birth defect or developmental disability. Some can be prevented. Others cannot. All of these people, however, need special care and treatment.