TERM	EXPLANATION/DEFINITION	NOTES
Cleft Lip or Palate	A child with cleft lip is born with part or the entire upper lip split or missing. In cleft palate, part or the entire roof (top) of the baby's mouth (inside) is missing at the time of birth.	
Heart Defect	The shape of the heart or connections of the blood vessels to the heart are not normal, and thus the heart does not move blood properly. The baby may look normal on the outside, but may also look blue, grey or pale all of the time or just with activity or feeding.	This can sometimes be seen on ultrasound, especially a special ultrasound called echocardiogram. It is not diagnosed just by looking at a baby or listening to the heart, although a heart murmur may be heard with some defects. Heart defects may cause severe weakness, turning blue while feeding or death.
Spina Bifida	A baby with spina bifida (also known as open spine) may have a sac sticking out of the middle or lower part of the back or the affected area may just be open with no skin or covering. The baby be unable to feel things normally (abnormal sensation) and to move the legs.	There may also be weakness or inability to move, or abnormal feet or legs.
Muscle Disease/ Muscular Dystrophy	The muscles are very weak and may get weaker over time. Sometimes, this may occur with other problems in the body, such as stomach problems, learning problems, etc.	This is diagnosed by a doctor getting a small sample of muscle out of the body for laboratory tests. In severe muscle disease, the person cannot walk.
Mental Retardation	The child does not learn like typical children. They learn slowly, may be unable to learn some things, and have difficulty doing things that are normal for children their age.	In severe mental retardation, the person may not be able to speak or control their bladder/bowels.
Down Syndrome	A child is born with a very small chin, weak muscles, a tongue that may rest outside of the mouth, a short neck, a very round face, and mental retardation (see above).	This is also called Trisomy 21. The child may also have difficulties with their digestion and heart defects.
Cystic Fibrosis	This disease causes difficulty breathing and frequent infections of the lungs. Children may have other problems in the body especially blockage of the bowels from thick mucus and not grow properly.	This is also sometimes called CF. Children may pass away at a young age. This disease can pass through families.
Kidney Disease	The body needs kidneys to help clean the blood and make urine. A person born with kidney disease may have problems growing, vomit often, have side or back pain, or may have no symptoms.	The first sign of a kidney problem in children may be high blood pressure; a low number of red blood cells, called anemia; or abnormal urine, including blood in the urine.
Sickle Cell Anaemia	This is a disease of the blood that causes people to have episodes of very severe pain, due to their blood not flowing properly through small veins. These painful episodes can be triggered by an infection.	The pain may be so bad that people need to go to the hospital for medicine. These children may be prescribed antibiotics and folic acid to take every day. This disease can pass through families.
Haemophilia	The body has difficulty controlling bleeding inside and from the skin. Even a small cut or minor surgery may cause a lot of bleeding. People with haemophila may have cuts that will start bleeding badly again after stopping at first.	Some people call this Bleeder's Disease. Sometimes, this is not diagnosed until a child has an accident of some sort. Many bruises may be on the skin. This disease can pass through families.
Thalassaemia	This is a disease of the blood that can cause fatigue. It is a type of anemia that can be mild or severe. It is not the common kind of anemia seen in pregnancy or in people who lack iron in their diet. The liver and spleen may be enlarged.	This is also sometimes called Mediterranean or Cooley's Anemia. It may be treated with blood transfusion. It can pass through families. It is more common in families that are Italian, Greek, Middle Eastern, Asian, or North African.

9/22/10





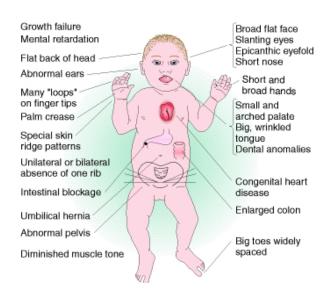


Cleft Lip Spina bifida

**People with Down syndrome** 



Muscular dystrophy



Some features of Down syndrome

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