

## BACKGROUND: GENETIC DISORDERS

**G**enetic disorders are conditions caused by abnormalities in the **genetic** material contained within our cells. There are more than 6,000 genetic disorders; some of which can lead to various health conditions. Children with genetic disorders may qualify for educational services or accommodations under a 504 Plan (Rehabilitation Act) or an Individualized Education Program (IEP) under the Individuals with Disabilities Education Act (IDEA). Criteria must be met within the school for these plans to be developed and implemented. For an overview of various genetic disorders, visit the Centers for Disease Control and Prevention and the National Human Genome Research Institute Web sites.

### Centers for Disease Control and Prevention

<http://www.cdc.gov>

### National Human Genome Research Institute

<https://www.genome.gov>

Below is an alphabetical list of some of the most common genetic disorders that an educator may see in the school setting:

**Becker muscular dystrophy** (BMD) is one of nine types of muscular dystrophy, a group of genetic, degenerative diseases primarily affecting voluntary muscles.

<http://www.mda.org>

**Bloom's syndrome** is caused by changes in a gene that is responsible for copying and repairing DNA in cells. These changes result in a high incidence of breaks in an individual's chromosomes. The most serious manifestations of this condition are a predisposition to cancer and a compromised immune system.

<https://www.jewishgenetics.org>

**Celiac disease** is an autoimmune disorder that can occur in genetically predisposed people where the ingestion of gluten leads to damage in the small intestine. It is estimated to affect 1 in 100 people worldwide. Two-and-one-half million Americans are undiagnosed and at risk for long-term health complications.

<http://celiac.org>

**Cystic Fibrosis** (CF) is a life-threatening genetic disease that primarily affects the lungs and digestive system. In people with CF, a defective gene and its protein product cause the body to produce unusually thick, sticky mucus.

[www.cff.org](http://www.cff.org)

**Down syndrome** occurs when an individual has a full or partial extra copy of chromosome 21. People with Down syndrome have an increased risk for certain medical conditions such as congenital heart defects, respiratory, and hearing problems. All people with Down syndrome experience cognitive delays.

[www.ndss.org](http://www.ndss.org)

**Duchenne Muscular Dystrophy (DMD)** is a genetic disorder characterized by progressive muscle degeneration and weakness. It is one of nine types of muscular dystrophy. DMD is caused by an absence of *dystrophin*, a protein that helps keep muscle cells intact. Symptom onset is in early childhood, usually between ages three and five. The disease primarily affects boys, but in rare cases it can affect girls.

[www.mda.org](http://www.mda.org)

**Fragile X syndrome (FXS)** is a genetic condition that causes intellectual disability, behavioral and learning challenges, and various physical characteristics. Though FXS occurs in both genders, males are more frequently affected than females, and generally with greater severity. Life expectancy is not affected.

<https://fragilex.org>

**Hemophilia A**, also called factor VIII (FVIII) deficiency or classic hemophilia, is a genetic disorder caused by missing or defective factor VIII, a clotting protein. People with hemophilia A often bleed longer than other people. Bleeds can occur internally, into joints and muscles, or externally, from minor cuts, dental procedures, or trauma.

[www.hemophilia.org](http://www.hemophilia.org)

**Huntington's disease (HD)** is a fatal genetic disorder that causes the progressive breakdown of nerve cells in the brain. It deteriorates a person's physical and mental abilities during their prime working years and has no cure. HD is known as the quintessential family disease because every child of a parent with HD has a 50/50 chance of carrying the faulty gene.

[www.hdsa.org](http://www.hdsa.org)

**Noonan syndrome** is a genetic disorder that prevents normal development in various parts of the body. A person can be affected by Noonan syndrome in various ways including unusual facial characteristics, short stature, heart defects, other physical problems, and possible developmental delays.

<http://www.mayoclinic.org>

**Sickle cell disease** is an inherited blood disorder that affects red blood cells. People with sickle cell disease have red blood cells that contain mostly hemoglobin S, an abnormal type of hemoglobin. Sometimes these red blood cells become sickle shaped (crescent shaped) and have difficulty passing through small blood vessels.

[www.sicklecelldisease.org](http://www.sicklecelldisease.org)

**Thalassemia** is an inherited blood disorder characterized by less hemoglobin and fewer red blood cells in the body than normal. This causes anemia, which frequently leads to fatigue.

<http://www.mayoclinic.org>

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