



# Genetic Diseases

A genetic disease is caused by the abnormal expression of one or more genes. These conditions can be caused by a number of possible pathways: genetic mutation, abnormal chromosome number, incorrect gene sequencing, and defective genes. Currently there are about 4,000 known genetic disorders, with new ones discovered every year. The vast majorities of the disorders are quite rare, and affect only one out of several thousand or million people. Cystic fibrosis is the most common genetic disorder, with approximately 5 percent of the U.S. population carrying a copy of the defective gene.

### Chronic Granulomatous Disease

Chronic granulomatous disease (CGD) is a diverse group of hereditary diseases in which immune system cells have difficulty forming reactive oxygen compounds necessary to kill ingested pathogens. This leads to the development of granulomas (a group of epithelioid macrophages surrounded by a lymphocyte cuff) in many organs that are ineffective at fighting infection. According to a study in 2004 by Maryland Pao, M.D. et al., CGD affects about 1 in 200,000 people in the United States, with at least 20 new cases diagnosed each year. Most cases of this disease are transmitted as a mutation on the X chromosome and are thus called an "X-linked trait." Symptoms include pneumonia, abscesses of the skin, tissues, and organs, arthritis, and superficial skin infections. Treatment revolves around early detection, education, and preventive antibiotics.

	COMPANY	PRODUCT	PHASE
Chronic Granulomatous Disease	InterMune, Inc.	Actimmune (interferon gamma-1b)	M

### Mucopolysaccharidosis

Mucopolysaccharidoses are a group of inherited metabolic diseases caused by the absence or malfunctioning of lysosomal enzymes needed to break down long chains of sugar carbohydrates that our cells use to build structures. These sugar carbohydrates (called glycosaminoglycans) are also found in the fluid found lubricating our joints. Because the large molecules are not broken down correctly, they accumulate in the cells, blood and connective tissues, causing permanent cellular damage. This affects the individual's appearance, physical abilities, organ system functions, and mental development. There are seven distinct clinical subtypes of the disease. There is currently no treatment for the disease but certain things are done to improve the quality of life. Physical therapy and exercise aid in movement, limiting dairy and sugar reduce mucus, and surgery to remove tonsils and adenoids aid in breathing.

	COMPANY	PRODUCT	PHASE
Mucopolysaccharidosis-VI	BioMarin Pharmaceutical, Inc.	Naglazyme / Aryplase (galsulfase)	M
Mucopolysaccharidosis-I	BioMarin Pharmaceutical, Inc.	Aldurazyme (laronidase)	PM

### Phenylketonuria

Phenylketonuria is a disorder in which the body lacks the enzyme necessary to metabolize phenylalanine to tyrosine. If not treated, the disorder can lead to brain damage and progressive mental retardation due to the excess build-up of phenylalanine in the body. The incidence of this disease varies widely, but averages about 1 in every 15,000 births. If the condition is diagnosed early, a child can grow up with normal brain development if a special diet is maintained. Phenylalanine must be severely restricted in the diet by limiting breast milk, meat, chicken, fish, nuts, cheese and other dairy products.

COMPANY	PRODUCT	PHASE	
<b>BioMarin Pharmaceutical, Inc.</b>	Phenoptin (sapropterin hydrochloride)	III	<b>Phenylketonuria</b>

### Turner's Syndrome

Turner syndrome encompasses several chromosomal abnormalities, with monosomy X being the most common. Instead of having two fully functional X chromosomes, only one is functional. Common symptoms of Turner include short stature, swelling of the hands and feet, broad chest, low hairline, low set ears, reproductive sterility, and several other individualized symptoms. On average, 98 percent of all fetuses with Turner syndrome spontaneously abort, and make up 10 percent of total spontaneous abortions. In the United States, Turner syndrome in live births is believed to be 1 in 2000.

COMPANY	PRODUCT	PHASE	
<b>Genentech, Inc.</b>	Nutropin AQ [somatropin (rDNA origin) injection]	M	<b>Turner's Syndrome</b>
<b>Genentech, Inc.</b>	Nutropin [somatropin (rDNA origin) for injection]	M	

