Genetic Disorders

Disorder	Cause	Description
Down syndrome (trisomy 21)	3 copies of chromosome 21 (nondisjunction during meiosis)	Mental retardation; increased birth defects; susceptible to many diseases
Turner's syndrome	Missing a copy of chromosome 23 (only one X present) – (nondisjunction)	Sterile female
Klinefelter's syndrome	3 copies of chromosome 23 (XXY) – (nondisjunction)	Sterile male
Tay-Sachs disease	2 recessive alleles on chromosome 15 (mutation)	Mental deficiency; blindness; death in early childhood
Sickle-cell anemia	2 codominant alleles on chromosome 11	Sickle-shaped red blood cells; blocked blood vessels; resistance to malaria
Cystic fibrosis	2 recessive alleles on chromosome 7	Digestive and respiratory problems (excess mucus in lungs); increased susceptibility to infections
Phenylketonuria (PKU)	2 recessive alleles on chromosome 12	Accumulation of phenylalanine in tissues; lack of normal skin pigment; mental retardation
Huntington's disease	1 dominant allele on chromosome 4	Mental deterioration; uncontrollable movements;
Hemophilia	Recessive allele on X chromosome (1 allele for males, 2 alleles for females)	Deficiency in blood clotting
Colorblindness	Recessive allele on X chromosome (1 allele for males, 2 alleles for females)	Inability to distinguish certain colors

Table 3-1 Some Birth Defects

Problem	Characteristics of Condition	Who Is at Risk	What Can Be Done
Alpha$_1$-antitrypsin deficiency	Enzyme deficiency that can lead to cirrhosis of the liver in early infancy and emphysema and degenerative lung disease in middle age.	1 in 1,000 white births	No treatment.
Alpha thalassemia	Severe anemia that reduces ability of the blood to carry oxygen; nearly all affected infants are stillborn or die soon after birth.	Primarily families of Malaysian, African, and Southeast Asian descent.	Frequent blood transfusions.
Beta thalassemia (Cooley's anemia)	Severe anemia resulting in weakness, fatigue, and frequent illness; usually fatal in adolescence or young adulthood.	Primarily families of Mediterranean descent	Frequent blood transfusions.
Cystic fibrosis	Body makes too much mucus, which collects in the lung and digestive tract; children do not grow normally and usually do not live beyond age 30; the most common inherited *lethal* defect among white people.	1 in 2,000 white births	Daily physical therapy to loosen mucus; antibiotics for lung infections; enzymes to improve digestion; gene therapy (in experimental stage).
Duchenne muscular dystrophy	Fatal disease usually found in males, marked by muscle weakness; minor mental retardation is common; respiratory failure and death usually occur in young adulthood.	1 in 3,000 to 5,000 male births	No treatment.
Hemophilia	Excessive bleeding, usually affecting males rather than females; in its most severe form, can lead to crippling arthritis in adulthood.	1 in 10,000 families with a history of hemophilia	Frequent transfusions of blood with clotting factors.
Neural-tube defects:			
Anencephaly	Absence of brain tissues; infants are stillborn or die soon after birth.	1 in 1,000	No treatment.
Spina bifida	Incompletely closed spinal canal, resulting in muscle weakness or paralysis and loss of bladder and bowel control; often accompanied by hydrocephalus, an accumulation of spinal fluid in the brain, which can lead to mental retardation.	1 in 1,000	Surgery to close spinal canal prevents further injury; shunt placed in brain drains excess fluid and prevents mental retardation.
Phenylketonuria (PKU)	Metabolic disorder resulting in mental retardation.	1 in 15,000 births	Special diet begun in first few weeks of life can offset mental retardation.
Polycystic kidney disease	*Infantile form;* enlarged kidneys, leading to respiratory problems and congestive heart failure.	1 in 1,000	Kidney transplants.
	Adult form; kidney pain, kidney stones, and hypertension resulting in chronic kidney failure.		
Sickle-cell anemia	Deformed, fragile red blood cells that can clog the blood vessels, depriving the body of oxygen; symptoms include severe pain, stunted growth, frequent infections, leg ulcers, gallstones, susceptibility to pneumonia, and stroke.	1 in 500 African Americans	Painkillers, transfusions for anemia and to prevent stroke, antibiotics for infections.
Tay-Sachs disease	Degenerative disease of the brain and nerve cells, resulting in death before age 5.	Historically found mainly in eastern European Jews	No treatment.

Source: Adapted from AAP Committee on Genetics, 1996; NIH Consensus Development Panel, 2001; Tisdale, 1988, pp 68–69.