TABLE 3.2

Common Genetic Diseases and Conditions

Name	Description	Prognosis	Probable Inheritance	Incidence*	Carrier Detection?†	Prenatal Detection?
Albinism	No melanin; person is very blond and pale	Normal, but must avoid sun damage	Recessive	Rare overall; 1 in 8 Hopi Indians is a carrier	No	No
Alzheimer's disease	Loss of memory and increasing mental impairment	Eventual death, often after years of dependency	Early onset—dominant; after age 60—multifactorial	Fewer than 1 in 100 middle-aged adults; perhaps 25 percent of all adults over age 85	Yes, for some genes; ApoE4 allele increases incidence	No
Breast cancer	Tumors in breast that can spread	With early treatment, most are cured; without it, death within 3 years	BRCA1 and BRCA2 genes seem dominant; other cases, multifactorial	1 woman in 8 (only 20 percent of breast cancer patients have BRCA1 or BRCA2)	Yes, for BRCA1 and BRCA2	No
Cleft palate, cleft lip	The two sides of the upper lip or palate are not joined	Correctable by surgery	Multifactorial	1 in every 700 births; more common in Asian Americans and American Indians	No	Yes
Club foot	The foot and ankle are twisted	Correctable by surgery	Multifactorial	1 in every 200 births; more common in boys	No	Yes
Cystic fibrosis	Mucous obstructions, especially in lungs and digestive organs	Most live to middle adulthood	Recessive gene; also spontaneous mutations	1 in 3,200; 1 in 25 European Americans is a carrier	Sometimes	Yes, in most cases
Diabetes	Abnormal sugar metabolism because of insufficient insulin	Early onset (type 1) fatal without insulin; for later onset (type 2), variable risks	Multifactorial; for later onset, body weight is significant	Type 1: 1 in 500 births; more common in American Indians and African Americans. Type 2: 1 adult in 6 by age 60	No	No
Deafness (congenital)	Inability to hear from birth on	Deaf children can learn sign language and live normally	Multifactorial; some forms are recessive	1 in 1,000 births; more common in people from Middle East	No	No
Hemophilia	Absence of clotting factor in blood	Death from internal bleeding; blood transfusions prevent damage	X-linked recessive; also spontaneous mutations	1 in 10,000 males; royal families of England, Russia, and Germany had it	Yes	Yes
Hydro-cephalus	Obstruction causes excess fluid in the brain	Brain damage and death; surgery can make normal life possible	Multifactorial	1 in every 100 births	No	Yes
Muscular dystrophy (30 diseases)	Weakening of muscles	Inability to walk, move; wasting away and sometimes death	Recessive or multifactorial	1 in every 3,500 males develops Duchenne's	Yes, for some forms	Yes, for some forms

*Incidence statistics vary from country to country; those given here are for the United States. All these diseases can occur in any ethnic group. Many affected groups limit transmission through genetic counseling; for example, the incidence of Tay-Sachs disease is declining because many Jewish young adults obtain testing and counseling before marriage.
†"Yes" refers to carrier detection. Family history can also reveal genetic risk.

Name	Description	Prognosis	Probable Inheritance	Incidence*	Carrier Detection?†	Prenatal Detection?
Neural-tube defects (open spine)	Anencephaly (parts of the brain missing) or spina bifida (lower spine not closed)	Anencephalic—severe retardation; spina bifida—poor lower body control	Multifactorial; folic acid deficit and genes	Anencephaly—1 in 1,000 births; spina bifida—3 in 1,000; more common in Welsh and Scots	No	Yes
Phenylketo-nuria (PKU)	Abnormal digestion of protein	Mental retardation, preventable by diet begun by 10 days after birth	Recessive	1 in 100 European Americans is a carrier, especially Norwegians and Irish	Yes	Yes
Pyloric stenosis	Overgrowth of muscle in intestine	Vomiting, loss of weight, eventual death; correctable by surgery	Multifactorial	1 male in 200, 1 female in 1,000; less common in African Americans	No	No
Rett syndrome	Neurological developmental disorder	Boys die at birth. At 6–18 months, girls lose communication and motor abilities	X-linked	1 in 10,000 female births	No	Sometimes
Schizophrenia	Severely distorted thought processes	No cure; drugs, hospitalization, psychotherapy ease symptoms	Multifactorial	1 in 100 people develop it by early adulthood	No	No
Sickle-cell anemia	Abnormal blood cells	Possible painful "crisis"; heart and kidney failure; treatable with drugs	Recessive	1 in 11 African Americans and 1 in 20 Latinos is a carrier	Yes	Yes
Tay-Sachs disease	Enzyme disease	Healthy infant becomes weaker, usually dying by age 5	Recessive	1 in 30 American Jews and 1 in 20 French Canadians and Old Order Amish are carriers	Yes	Yes
Thalassemia	Abnormal blood cells	Paleness and listlessness, low resistance to infections, slow growth	Usually recessive, occasionally dominant	1 in 10 Americans from southern Europe, northern Africa, or south Asia is a carrier	Yes	Yes
Tourette syndrome	Uncontrollable tics, body jerking, verbal outbursts	Appears at about age 5; worsens then improves with age	Dominant, but variable penetrance	1 in 250 children	Sometimes	No

Sources: Briley & Sulser, 2001; Butler & Meaney, 2005; Klug & Cummings, 2000; Mange & Mange, 1999; K. L. Moore & Persaud, 2003; Shahin et al., 2002.

Observation Quiz (see answer, page 84): Is there any ethnic group that does not have a genetic condition that is more common among its members than among the general population?