Genetic Testing Timeline

The histories of the most well-known single-gene disorders started long before anyone ever thought about sequencing the human genome (see story p. 24).

**Huntington Disease**

- **1840s** Medical journals note involuntary movements and mental problems that run in families
- **1872** At 22-years-old, physician George Huntington publishes a paper describing symptoms and hereditary pattern of HD, based on observations of his father's patients
- **1983** HD marker discovered
- **1986** First predictive test for HD
- **1990** Psychological and safety assessment for HD testing begins
- **1993** Huntington gene discovered at 4q16.3

**Cystic Fibrosis**

- **1938** Dorothy Andersen describes disease as defect in exocrine gland ducts
- **1951** "Salty sweat" discovered in infants with heat prostration in New York City, confirming 17th-century rhyme about children "salty to taste" who do not live long
- **1986** Several research groups identify linked markers on chromosome 7
- **1989** CFTR gene discovered at 7q31.2
- **2001** NIH, the American College of Obstetricians and Gynecologists, and the American College of Medical Genetics recommend screening before and during pregnancy

**Sickle Cell Disease**

- **1904** Medical intern Ernest Irons notices "pear-shaped and elongated forms" in blood from Clement Noel, a West Indian dental student in Chicago
- **1910** James Herrick, Irons' attending physician, takes credit for discovery
- **1958** Vernon Ingram and coworkers use peptide fingerprinting to localize a mutation at the sixth amino acid position of β globin
- **1972** Congress passes National Sickle Cell Anemia Control Act establishing carrier screening; Air Force Academy excludes SCD carriers
- **1981** Air Force Academy stops excluding SCD carriers
- **1982** Prenatal diagnosis of SCD by direct gene test at 11p15.5
- **1985** SCD carrier status dubbed risk factor for sudden death during intense physical training or high-altitude exposure
- **1987** NIH consensus statement recommends newborn screening for SCD

**Tay-Sachs Disease**

- **1881** British ophthalmologist Warren Tay describes "cherry red spot"
- **1887** US neurologist Barnard Sachs describes neurological symptoms
- **1983** Josef Ekstein starts Dor Yeshorim, an organization to screen for "Jewish genetic diseases," after he loses four children to TSD
- **1987** hexA gene and mutations described at 15q23-q24
- **2003** Dor Yeshorim has screened 100,000+ young people for a dozen Jewish genetic diseases since its inception. Data stored anonymously, consulted for marriage decisions

—Compiled by Ricki Lewis