

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

1981
Nancy Wexler begins field studies on Venezuelan families, as others establish linkage on chromosome 4

1983
HD marker discovered

1986
First predictive test for HD

1990
Psychological and safety assessment for HD testing begins

1993
Huntingtin gene discovered at 4p16.3



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

1997
A 10-year study shows newborn screening for CF, early dietary intervention, and antibiotics improve health; NIH recommends prenatal screening

2001
NIH, the American College of Obstetricians and Gynecologists, and the American College of Medical Genetics recommend screening before and during pregnancy



Illustrations: Thom Graves Media

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

1970
Carrier screening begins in Maryland, quickly becomes global

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