

OUR GENES: WHAT THE MAP SHOWS

Neurofibromatosis, Type 2 ◉
Tumors of the auditory nerves and tissues surrounding the brain.

Duane's Syndrome ◉
Congenital mental deficiency condition marked
Three copies of chromosome 21

Amyotrophic Lateral Sclerosis* ◉
(Lou Gehrig's Disease)
Fatal degenerative nerve ailment

ADA Deficiency ◉
Severe susceptibility to infections.
First hereditary condition treated
by gene therapy

Familial Hypercholesterolemia ◉
Extremely high cholesterol

Myotonic Dystrophy ◉
Frequent form of adult
muscular dystrophy

Amyloidosis ◉
Accumulation in the
tissues of an insoluble
fibular protein

Brain Cancer* ◉
5% to 10% of cases

Polycystic Kidney Disease ◉
Cysts resulting in enlarged kidneys
and renal failure

Hemophilia ◉
Blood defect
making it difficult
to control
hemorrhaging

Muscular Dystrophy ◉
(Duchenne and Becker types)
Progressive deterioration of the muscles

ALD ◉ (adrenoleukodystrophy)
Nerve disease portrayed in movie
Lorenzo's Oil

Cocher's Blotches ◉
A chronic pigment
deficiency occurring
frequently among
Ashkenazi Jews

Familial Colon Cancer* ◉
One in 200 people have this gene;
of those, 65% are likely to
develop the disease

Retinitis Pigmentosa* ◉
Progressive degeneration of the retina

Huntington's Disease ◉
Neurodegenerative disorder tend to
strike people in their 40s and 50s

**Familial Polyposis
of the Colon** ◉
Abnormal tissue growth
frequently leading to cancer

Hemochromatosis ◉
Abnormally high
absorption of iron from
the diet

Spinocerebellar Ataxia ◉
Destroys nerves in the
brain and spinal cord
resulting in loss of
muscle control

Cystic Fibrosis ◉
Mucus fills up the
lungs, interfering with
breathing. One of the
most prevalent
genetic diseases
in the U.S.

Multiple Exostoses* ◉
A disorder of cartilage
and bone

Malignant Melanoma ◉
Tumors originating in the skin

Multiple Endocrine Neoplasia, Type 2 ◉
Tumors in endocrine glands and other tissues

Sickle Cell Anemia ◉
Chronic inherited anemia, primarily affecting blacks, in
which red blood cells sickle, or form crescents, plugging
arteries and capillaries

Alzheimer's Disease* ◉
Degenerative
nerve disease
marked by
prevalence
usually

Retinitis Pigmentosa* ◉
A relatively common tumor
of the eye, accounting for 2% of
childhood malignancies

PKU ◉
(phenylketonuria)
An inborn error of metabolism
that frequently results in mental retardation

WHEN CAN WE GET TESTED?

- ◉ DNA test currently available
- ◉ Gene mapped but not yet isolated
- ◉ Diagnosis available through family-linkage study of DNA markers
- ◉ DNA test under development

* One form of the disease

HOW MANY PARTS TO THE MICROSCOPIC PARTS

The human body contains 100 trillion cells



There is a small nucleus inside each human cell



Each nucleus contains 46 chromosomes, arranged in 23 pairs



One chromosome of every pair is from each parent



The chromosomes are filled with tightly coiled strands of DNA



Genes are segments of DNA that contain instructions to make proteins—the building blocks of life



Tay-Sachs Disease ◉
Fatal hereditary disorder involving lipid metabolism often occurring in Ashkenazi Jews and French Canadians

THIS GRAPH BY THE U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES, NATIONAL INSTITUTES OF HEALTH, SOURCE: BY PETER A. MARRAS, JOURNAL PUBLISHERS, INC.