

What Can We Learn From Genetic and Genomic Testing

Jonathan Clyman, PhD.
Board certified Genetic Counselor
Dyson Cancer Center

HEALTHQUEST

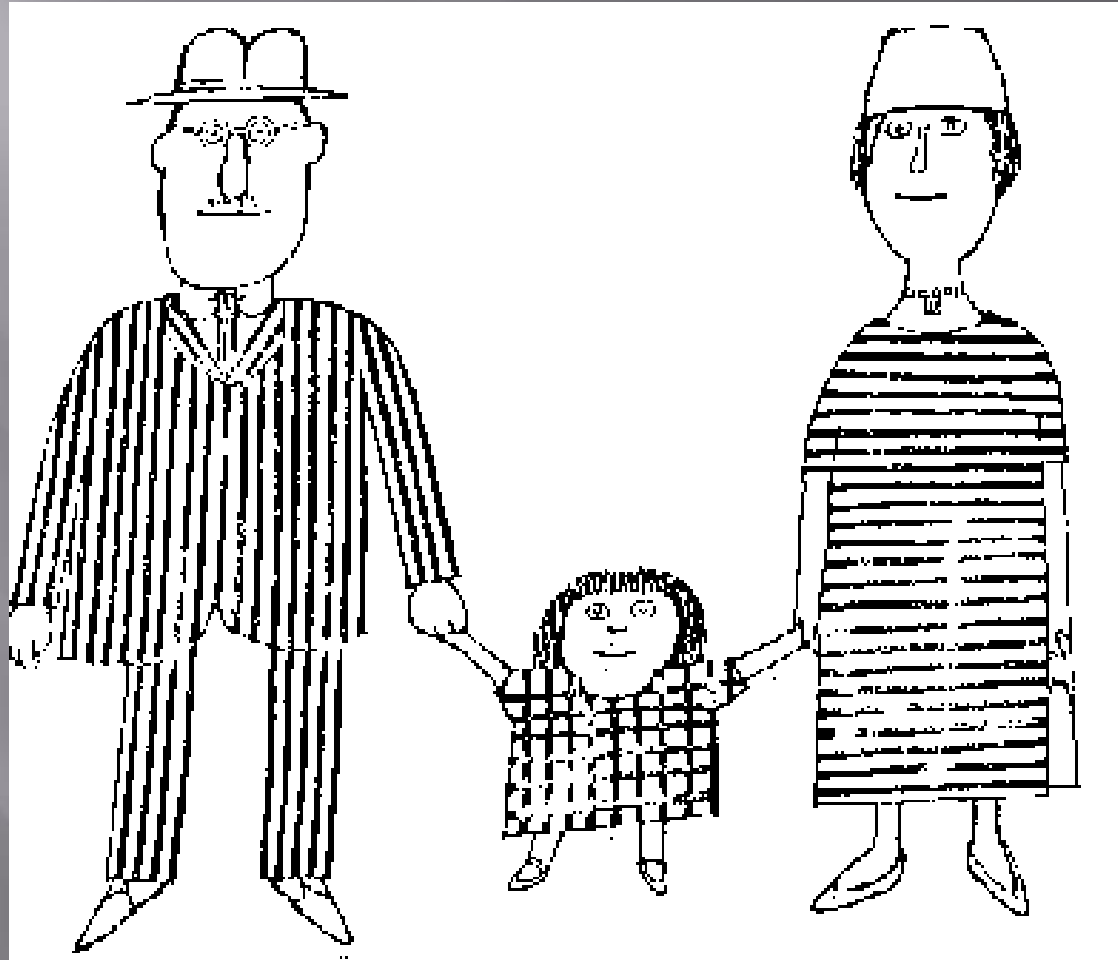
GENETICS vs GENOMICS

- Genetics is the study of inherited traits and genes
- Genomics is the study of how a set of genes behave

GENETIC vs. GENOMIC



Some basic genetics



Some basic genetics

Chromosomes



Nucleus

Chromosome

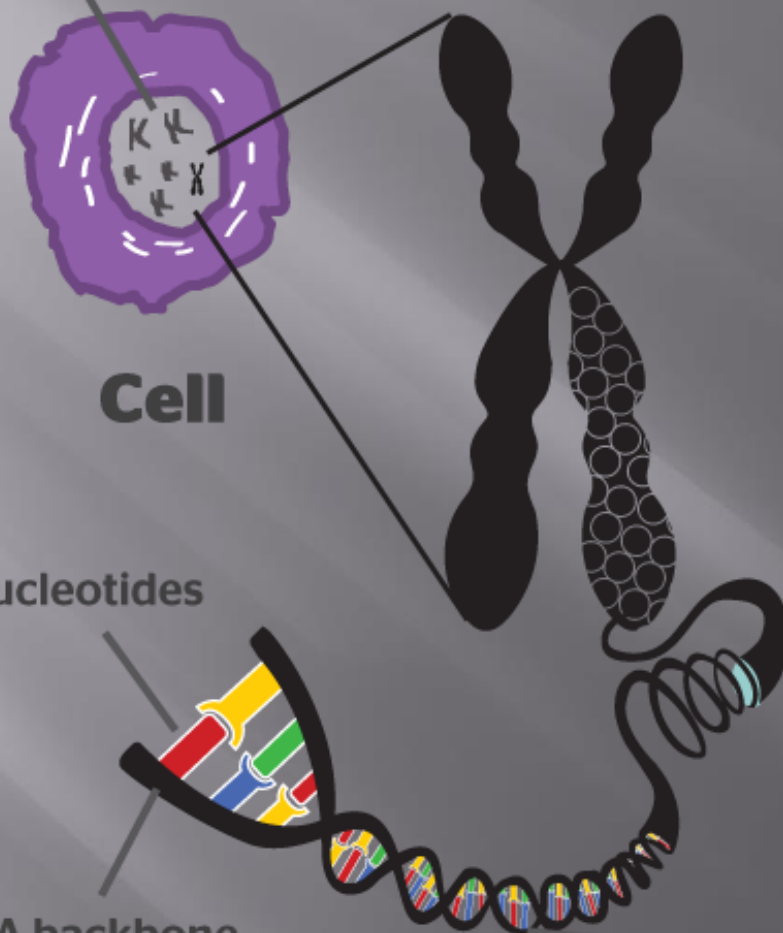
Cell

Nucleotides

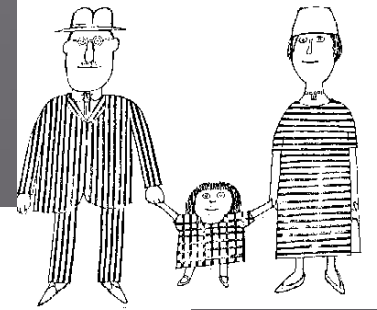
DNA backbone

Coiled DNA molecule

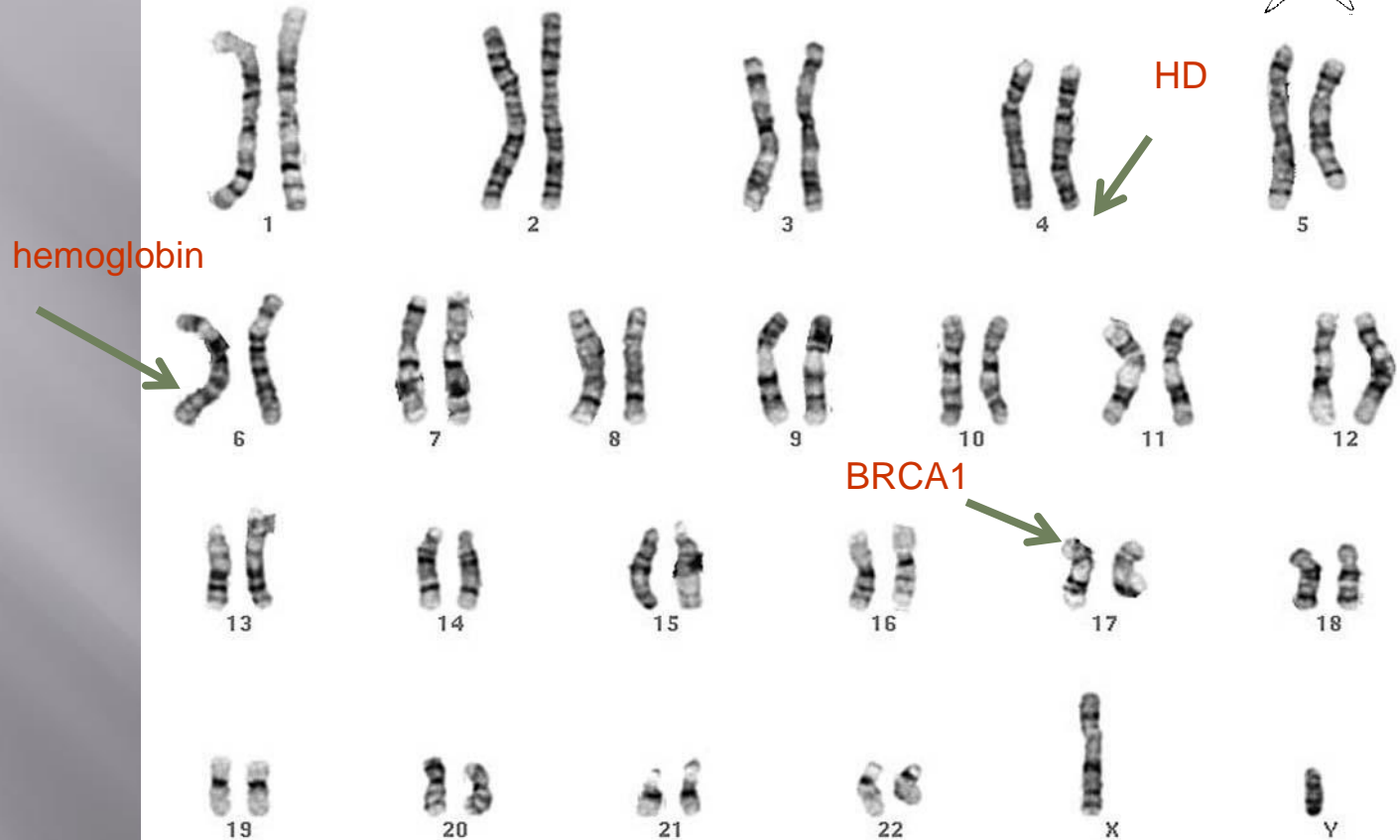
DNA double helix



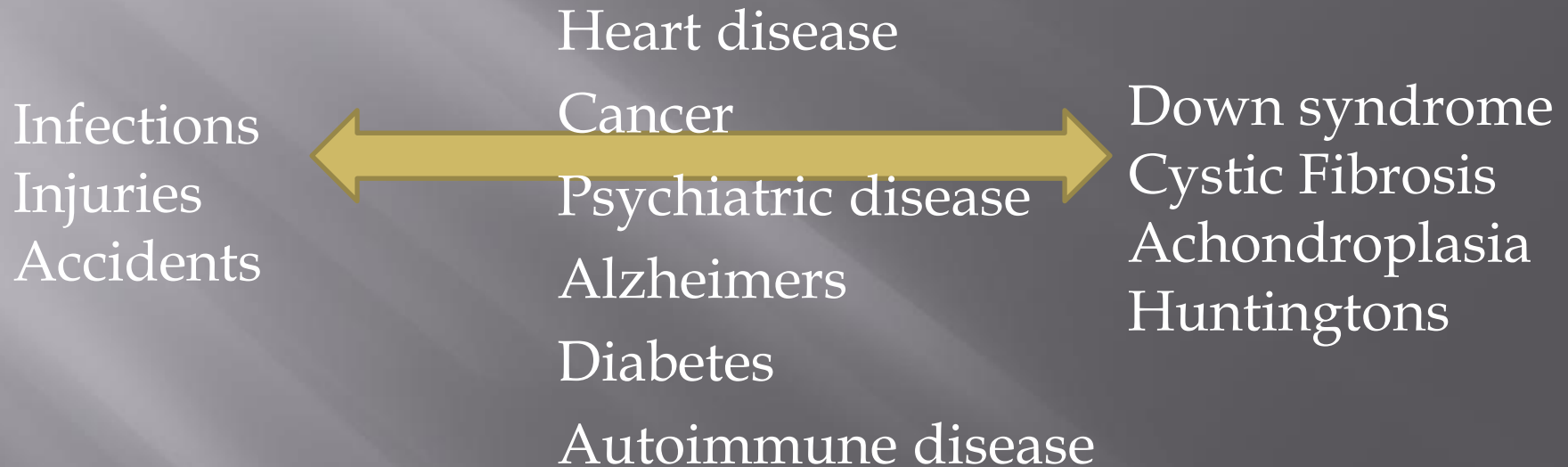
23 pairs of chromosomes



Human male
G-bands



Genes and Disease



Breast Cancer Genetics

- ALL cancer is genetic ...
 - Complex, multifactorial etiology

BREAST CANCER GENETICS

- ALL cancer is genetic ...

BUT

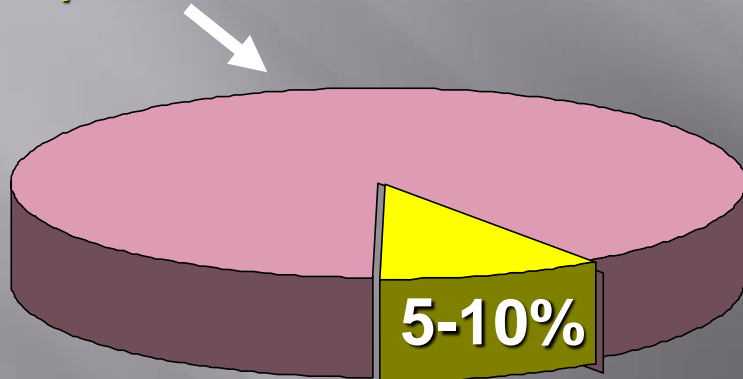
- GENETIC is not the same as INHERITED
 - Only a small portion of cancer is inherited

Hereditary Breast Cancer

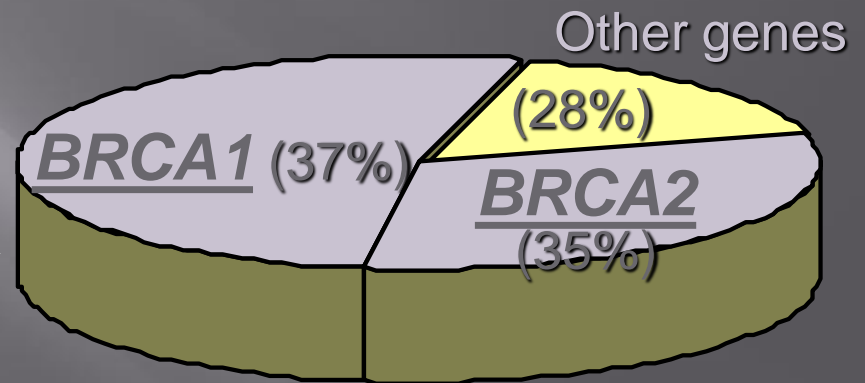
Complex genetics
and environmental factors

90-95%

Sporadic/Familial



Hereditary



The BRCA1 and BRCA2 genes are
associated with most inherited breast cancer

Am J Hum Genet 1998; 62:676-89

J Clin Onc 2002;20:1480-1490

J Clin Onc 2007, 25; 6-7

***BRCA* mutation cancer risks**

	General Population	<i>BRCA1/BRCA2</i> mutation
Breast	12%	40-87%
Ovarian	1-2%	11-40%
Male Breast	0.1%	1-10%
Prostate	15-18%	<30-39%
Pancreatic	0.5%	1-7%

Increased risk for a second primary breast cancer

Reports of increased risk for melanoma

What can we learn from genetic testing?

For a woman diagnosed with breast cancer -

- Can affect surgical decisions and risk management
- If a BRCA mutation is identified, other family members can be tested

If BRCA testing is negative...

What can we learn from genetic testing?

For women AT HIGH RISK for breast cancer

- ▣ Can affect risk management
 - Increased screening
 - Chemoprevention
 - Preventative surgery

If BRCA testing is negative...

GENETIC vs. GENOMIC

