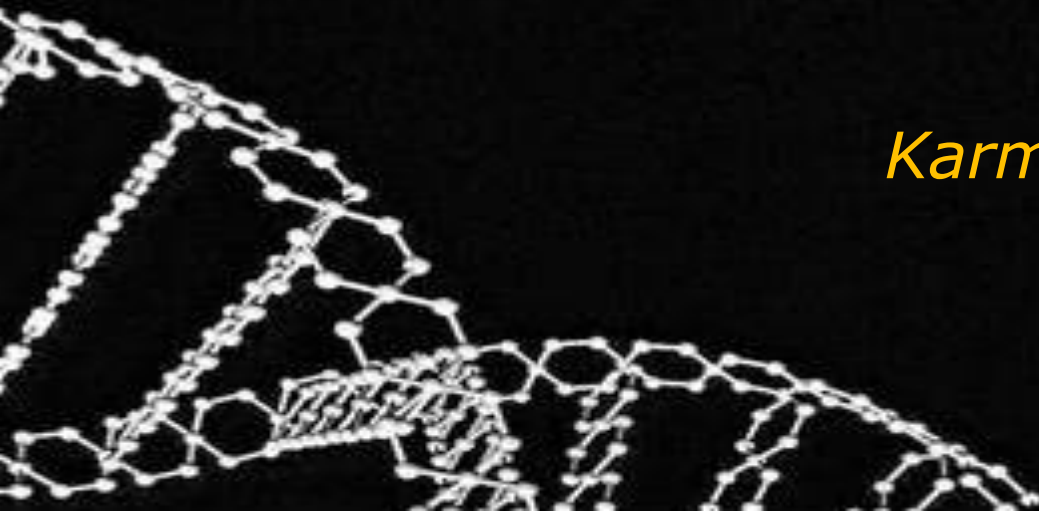
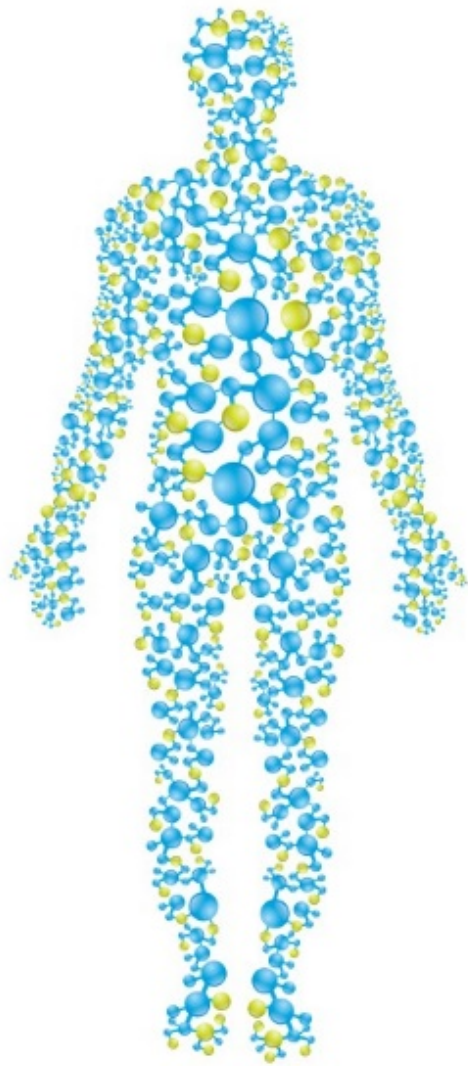


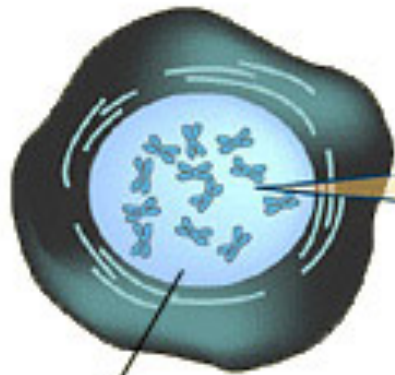
USH Talks

# Genetics 101

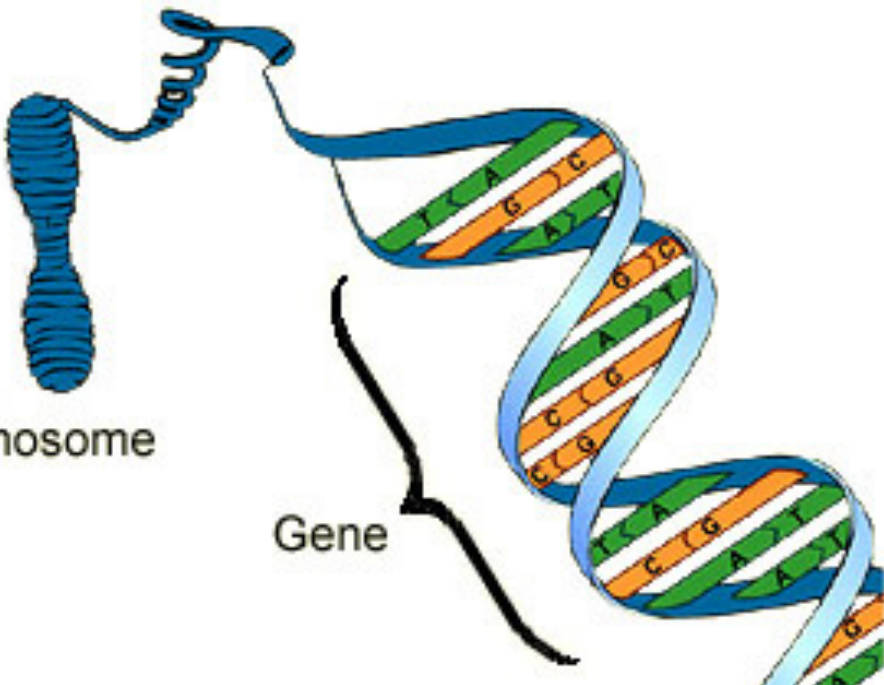
*Karmen M. Trzupek, MS, CGC  
Genetic Counselor*








Nucleus



Chromosome

Gene



Why the long face?

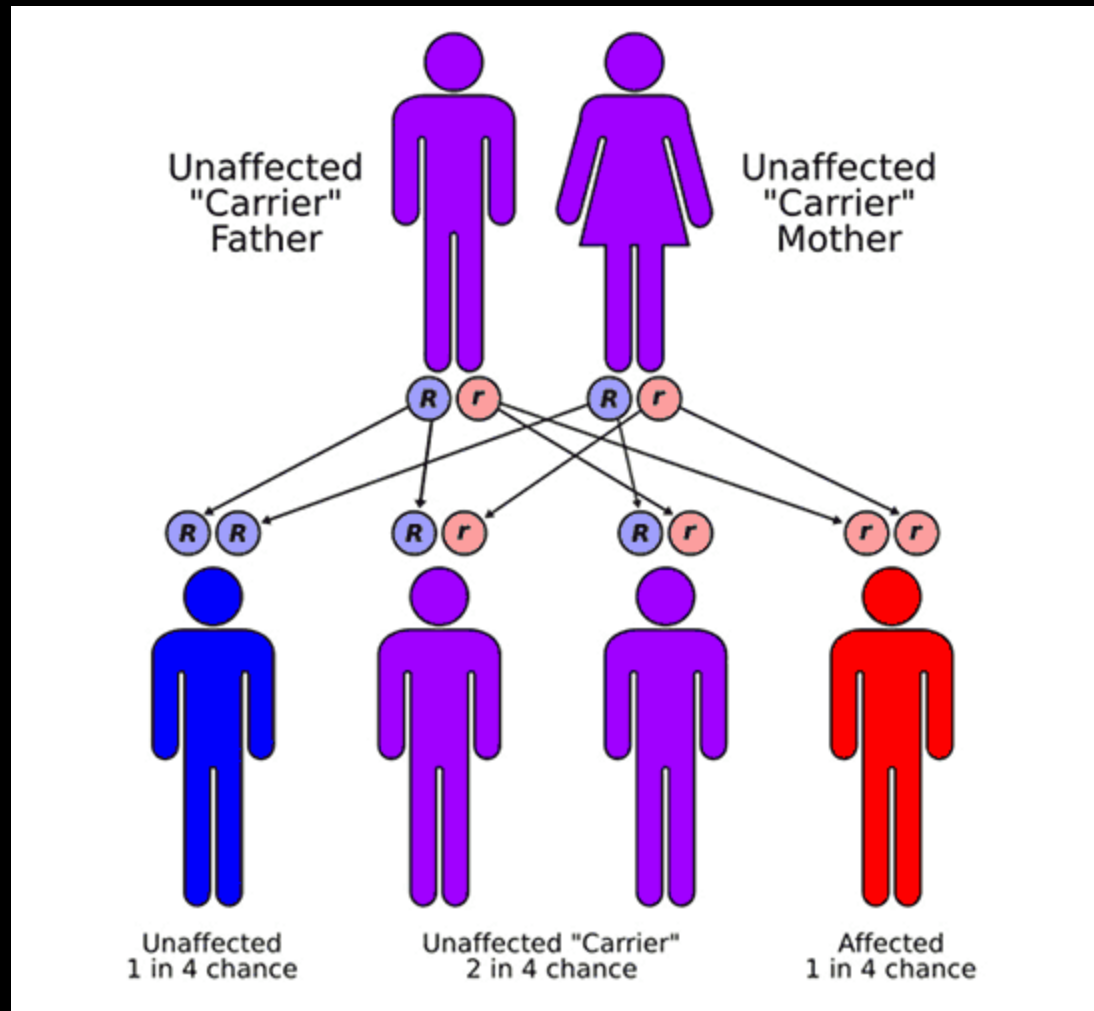
heredity.

# Chromosomes

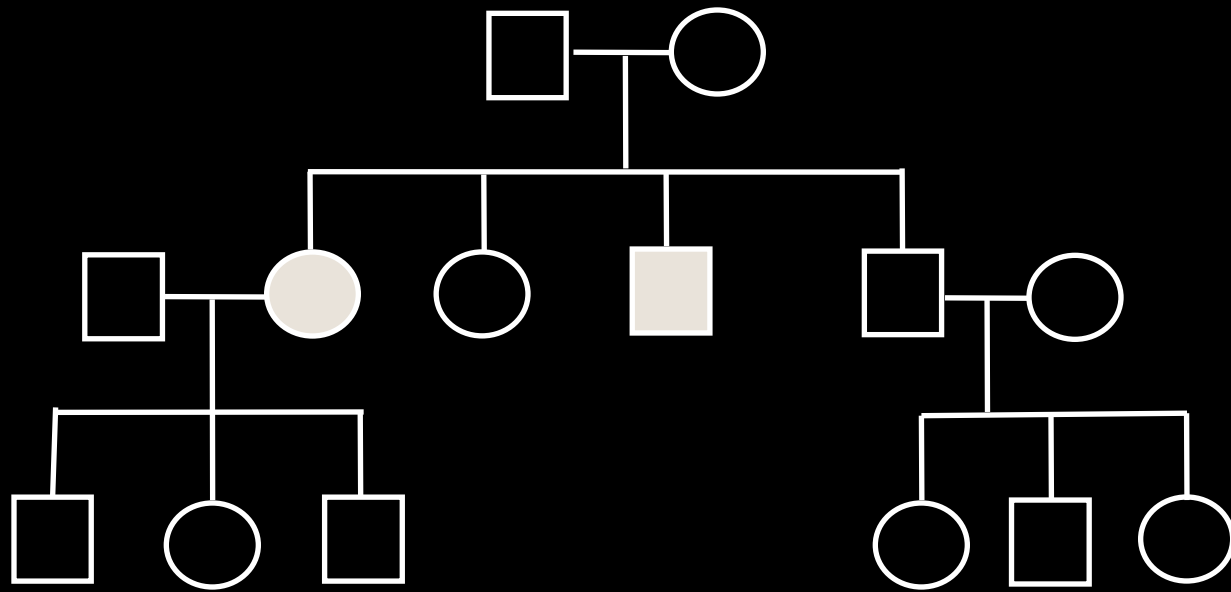
Normal female: 46,XX



# Usher syndrome Inheritance: Autosomal Recessive



# Recessive Inheritance

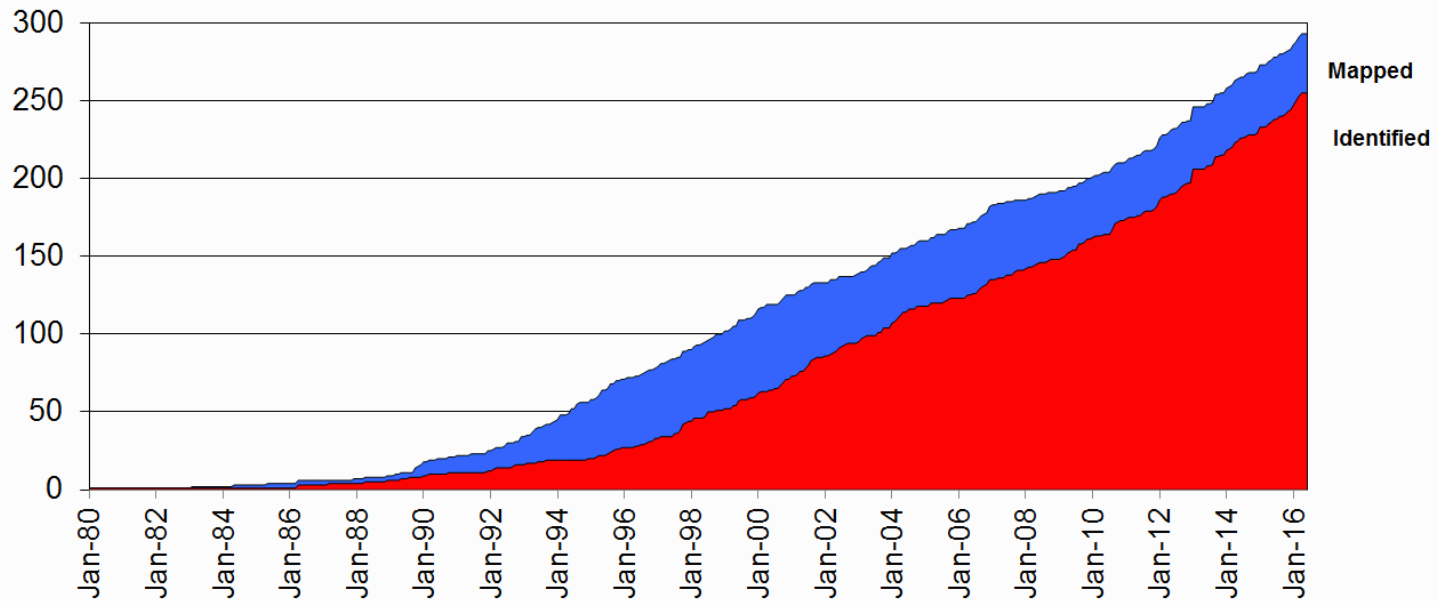






# Usher syndrome genes

Total # of Mapped Genes	Identified Genes
15	11+

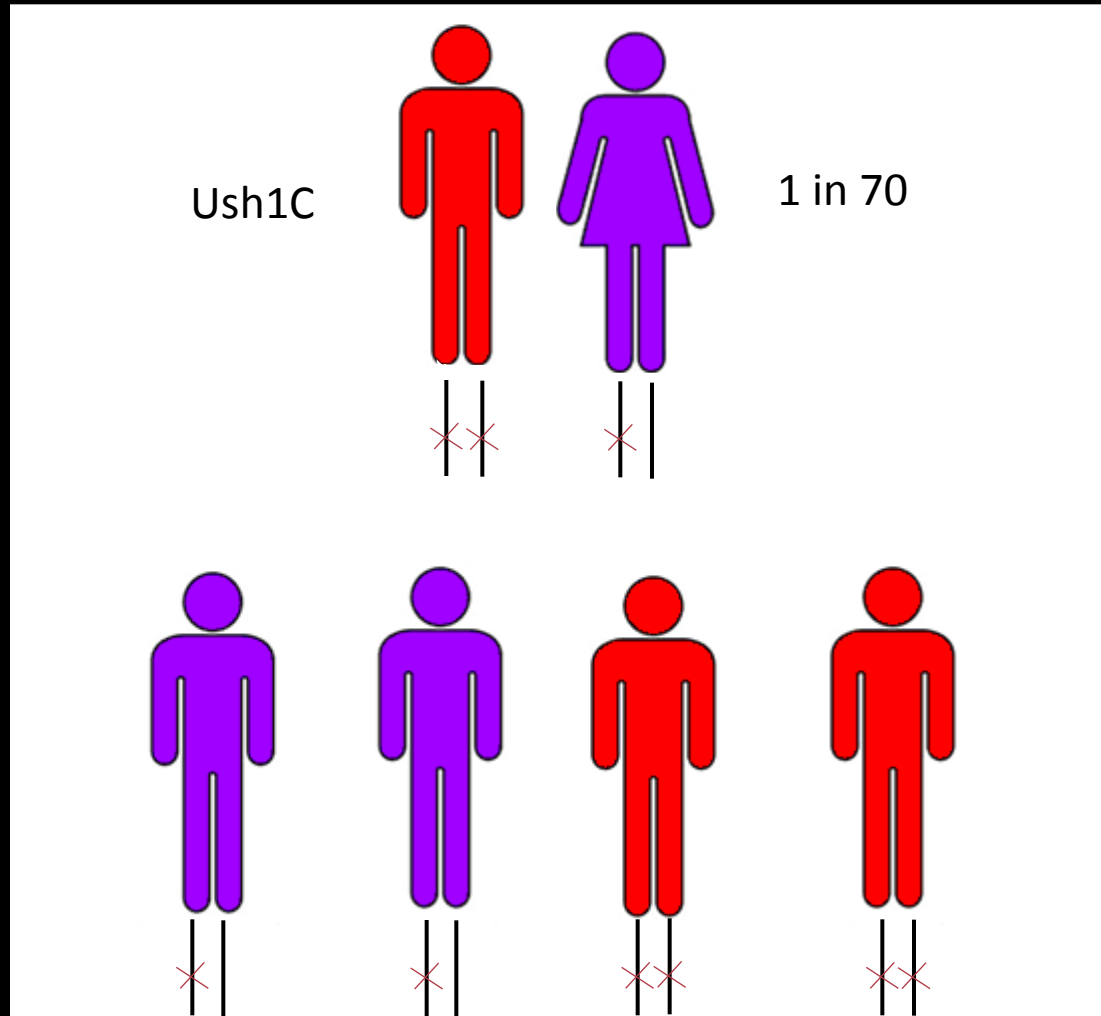


**Mapped and Identified Retinal Disease Genes 1980 - 2016**

# Usher syndrome type 1

Usher syndrome subtype	Gene Name	Relative Incidence
USH1B	<i>MYO7A</i>	50-60%
USH1C	<i>Ush1C</i>	6-7%
USH1D	<i>CDH23</i>	15-20%
USH1E	?	Rare
USH1F	<i>PCDH15</i>	10-15%
USH1G	<i>SANS</i>	~7%
USH1H	?	?
USH1J	<i>CIB2</i>	?
USH1K	?	?

# Ush1C in Louisiana



# Usher syndrome types 2 & 3

Usher syndrome subtype	Gene Name	Relative Incidence
USH2A	<i>Ush2A</i>	~80%
USH2C	<i>VLGR1 (GPR98)</i>	~15%
USH2D	<i>WHRN</i>	~5%
USH1E	<i>Unknown</i>	Rare

Usher syndrome subtype	Gene Name	Relative Incidence
USH3A	<i>CLRN1</i>	>95%
USH3B	<i>HARS</i>	Rare



USHER SYNDROME  
COALITION

The logo for InformedDNA features a stylized 'i' in blue with a green dot above it. The word 'INFORMED' is in blue, 'DNA' is in green, and the tagline 'Healthcare, Personalized.' is in green below it.

INFORMEDDNA  
Healthcare, *Personalized.*