Genetic Research

Basic Vocabulary and Terminology

Bertha deLanda
Research Compliance Office
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http://video.nytimes.com/video/2010/04/21/us/1247467672743/blood-journey.html

Grand Canyon, Arizona

Carletta Tilousi, Havasupai Tribal Council

Population Genetics - a branch of genetics concerned with gene frequencies and genotype frequencies in populations

Began by testing for diabetes, studies expanded to other institutions to include:

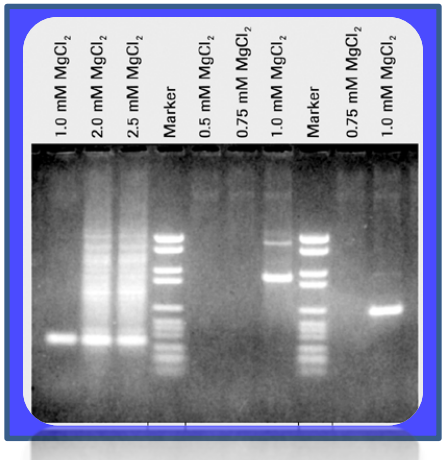
- alcoholism
- schizophrenia
- in-breeding
- origin of the tribe

Information

Criteria for Approval 45 CFR 46.111 [OHRP]

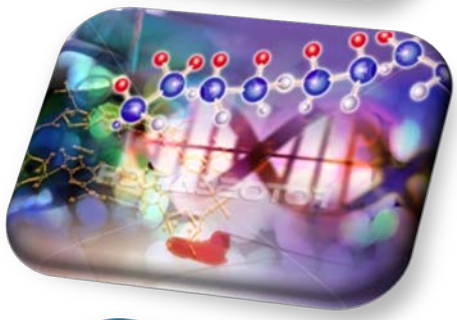
- Privacy of subjects and confidentiality of the data is protected.

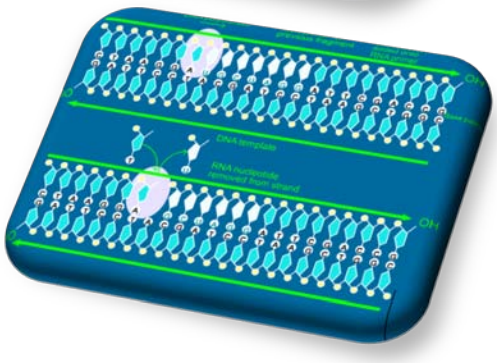
- By researching DNA, scientists can collect information about an individual's:



  - **Identity** (sex, ethnic background, forensics, paternity)
  - **Genetic disorders** (Fragile X, Huntington's Disease)
  - **Genetic predisposition** to future diseases (HPV, cancer)
  - **Hereditary traits** that can be passed on to offspring (Sickle Cell anemia, color blindness, Tay Sachs Disease)

Today's Presentation





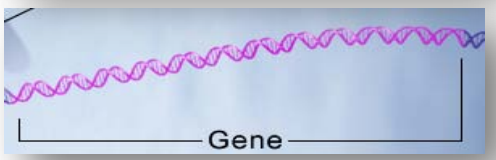
- **Clarify/define** a few basic terms and vocabulary used in genetic research
- **Provide vocabulary list** to help reviewers identify a study as possibly including genetic testing
- **Emphasize goal** : Not to learn/memorize the terms and words, but to help reviewers recognize when studies involve genetic testing

Researchers may include genetic testing as part of their studies, without considering it **genetic research**

Definitions: in Lay Language

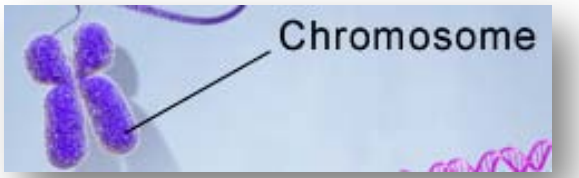
DNA – strand of genetic information

Gene – hereditary unit of DNA

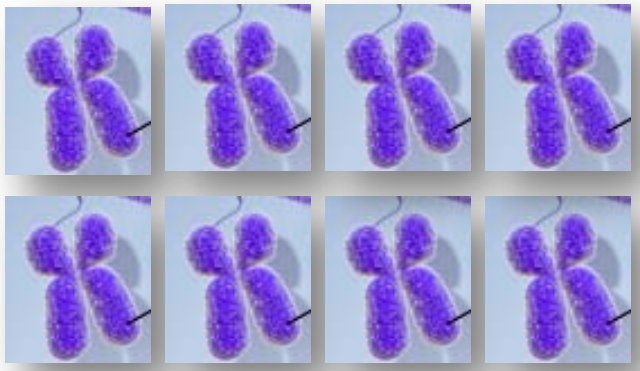


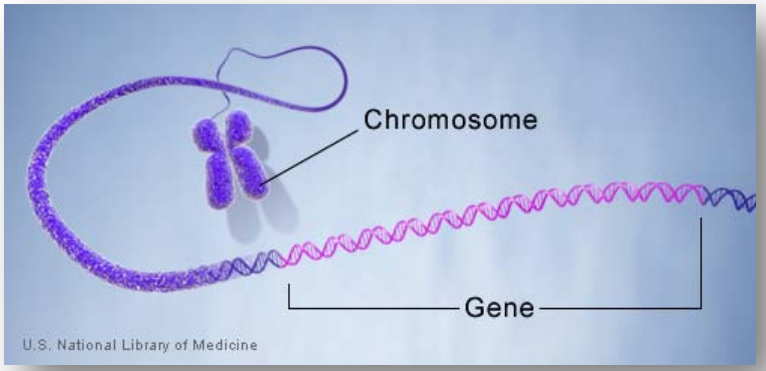
Chromosome – carry the genes in a linear order (humans have 23 pairs)

Genome – full set of chromosomes



Whole Genomic Sequencing – entire decoding of someone's DNA





Terms: In Lay Language

Biomarker
Allele
Loci
SNPs
STRs

Terms that indicate a location or mutation found on a DNA sequence

Personal Genomics
Personalized medicine

Pharmacogenetics/ Pharmacogenomics

Epigenomics/ Epigenetics

Involve modifying specified DNA targets to see how information in genes is expressed and used by other cells

To optimize drug therapy, with respect to the patients' genotype, to ensure maximum efficacy with minimal adverse effects

Genotype versus Phenotype

- **Phenotype**: visible trait
- **Genotype** : actual genetic sentence

For example:

Phenotype:	Brown Hair	Brown Hair	Blonde Hair
Genotype:	Brb Recessive blonde	BrBr Dominant brown	Brb Recessive blonde

Genotype versus Phenotype

Genotypic behavior versus **phenotypic behavior**:
Three people have the genetic traits for OCD; one washes their hands constantly, one feels the need to obsess about brushing their teeth and one may feel the need to hoard items

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Genotype:	Brb Recessive blonde	BrBr Dominant brown	Brb Recessive blonde

GINA :

Genetic Information Nondiscrimination Act

Signed by President Bush in May 2008

(GINA)…will protect Americans against discrimination based on their genetic information when it comes to health insurance (Title I) and employment (Title II)

"The long-awaited measure, which has been debated in Congress for 13 years, will pave the way for people to take full advantage of the promise of personalized medicine without fear of discrimination."

When reviewing a protocol the IRB should:

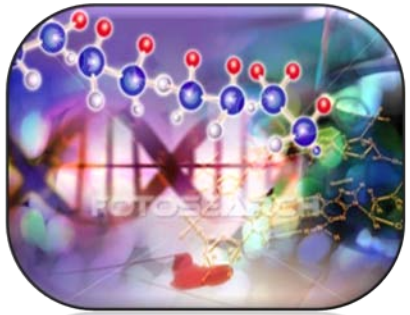
- Try to **determine** if genetic testing is part of the study, if necessary

- Determine whether or not the **information is necessary** to achieve study goal
- Ensure proper **safeguards** are in place

- Check for **GINA language** in consent:

"…A Federal law, the Genetic Information Nondiscrimination Act of 2008 (GINA), generally makes it illegal…to discriminate against you based on your genetic information.

- Ensure the subject is adequately informed about what will happen to their samples/data



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- The IRB receives and reviews the relevant information to evaluate research studies *(AAHRPP Element II.2.D)*
- Risks to subjects are minimized *(45 CFR 46.111(a)(1)*
- Research plan makes adequate provisions for monitoring the data collection to ensure subject safety *(45 CFR 46.111(a)(6)*
- No informed consent may include… exculpatory language *(45 CFR 46.116)* and
- The information given to the subject shall be in language understandable to the subject *(45 CFR 46.116)*