# GENOMICS IN SOCIETY

## GWAS – Genome-Wide Association Studies

# GWAS – what is it?

**Genome-Wide Association Study =**

**Genome** → complete set of genes or genetic material present in a cell or organism

**Wide** → including the full extent & variety of the genome being studied

**Association** → when one or more genotypes within a population co-occur with a phenotypic trait more often than would be expected by chance occurrence

**Study** → a detailed investigation & analysis used to answer a research question

# GWAS – what is it?

**Therefore…**

Genome-Wide Association Studies look at the entire genome of an organism of interest all at once to detect specific genotypes and determine if those genotypes are associated with specific phenotypes such that researchers can answer whether or not a trait is genetic.

# GWAS – technical definitions

"A genome-wide association study (GWAS) is an approach used in genetics research to associate specific genetic variations with particular diseases." – National Human Genome Research Institute

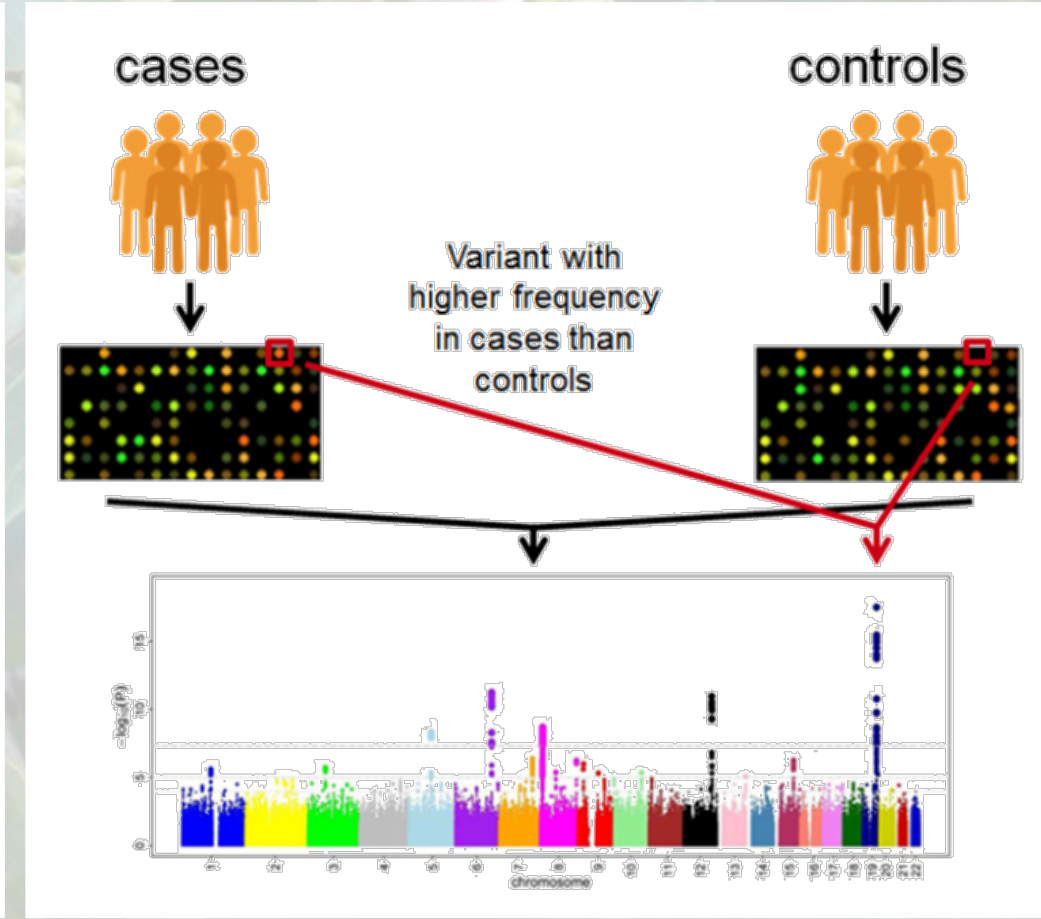
"Genome-wide association studies (GWAS) involve testing genetic variants across the genomes of many individuals to identify genotype–phenotype associations." – Nature Research Journal

"A genome-wide association study (GWAS) is a new approach that involves rapidly scanning several hundred thousand (up to 5 millions) markers across the complete sets of DNA of many people to find genetic variations associated with a particular trait." – ScienceDirect

"Genome wide association studies (GWAS) are hypothesis-free methods for identifing associations between genetic regions (loci) and traits (including diseases)." – GWAS Catalog

# GWAS – how does it work?

- Identify many individuals (peoples, trees, etc.) that have detectable differences (different traits/phenotypes)
- Search across the entire genome for base-pair differences, i.e. where the genotypes have different A/C/G/T
- Try to find where there is a consistent difference in the genome – where are the base pairs consistently different when a specific trait is present?
- If successful, able to focus on a place/region in the genome that must be involved in this difference
- Don’t have to guess ahead of time what kind of gene you're trying to find/where the gene is in the genome



# GWAS – when was it first used?

- 2002: first successful study conducted with human genome
  - Led to landmark 2005 study that found very strong genetic association to a human disease
- **Thousands** of studies have been conducted to date
  - Many more studies to come as the technology advances

*It is a relatively 'new' technology in scientific research*

# GWAS – who uses it?

- Can be used whenever a scientist is trying to find the different genotypes within an entire genome that are associated with phenotypes
- Used in many different types of genetics research, such as:
  - Medicine
  - Forestry
  - Agriculture
  - Wildlife

# GWAS – why is it used?

- Used to look for variation in an entire genome instead of looking at small, pre-determined portions a genome
- It allows scientists to uncover more consistent genetic trends throughout a study population
- Scientists can see the full range of variation in a genome instead of trying to piece together genomic segments

# GWAS – pros & cons

## PROS

- No need for candidate gene – able to say the whole set of genes are candidates & consider all of them, and have a strategy that's comprehensive enough to do that
- Do not have to assemble fragments of a genome, can see the full range of genetic variation at once
- Provides insight into genetically associated traits that then allows scientists to better manage these traits or select these traits

## CONS

- Associations are sometimes very weak
- 'Newer' technology, many more advances to be made still to improves its accuracy & efficiency
- Has to be done using advanced machines & computers, located at specific laboratories
- Time consuming process
- Expensive—costs a lot of money for scientists to conduct these studies, and sometimes the results are not conclusive

# GWAS – summary

**Overall…**

GWAS are useful to many scientists who study different types of living organisms because they are able to look at a genome all at once without having to know what they are looking for beforehand. Scientists can see a lot of different genetic variation and then determine what traits are associated with those variants. Scientific technology is continually advancing, so while there are pros & cons to GWAS, it has been useful in discovering a lot of genetic variation over the close to 20 years it has been being used.