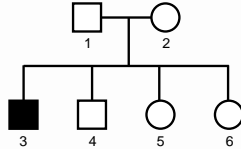




Human Genetic Diseases



Pedigree analysis

- Pedigree analysis reveals Mendelian patterns in human inheritance
- data mapped on a family tree

□ = male ○ = female ■ = male w/ trait ● = female w/ trait

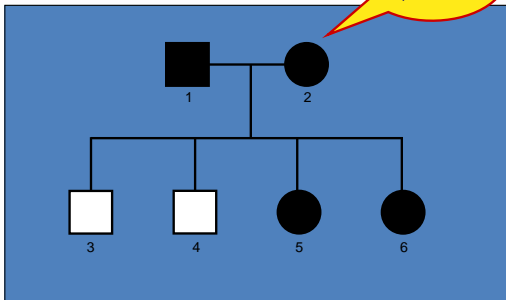
Widow's peak
 First generation: $Ww \times ww$
 Second generation: Ww, ww, Ww, ww
 Third generation (two sisters): WW or Ww

Attached earlobe
 First generation: $Ff \times Ff$
 Second generation: FF or Ff, ff
 Third generation: Ff or ff

Free earlobe
 First generation: $Ff \times Ff$
 Second generation: Ff, ff
 Third generation: Ff or ff

Simple pedigree analysis

What's the likely inheritance pattern?



AP Biology

Tay-Sachs (recessive)

- Primarily Jews of eastern European (Ashkenazi) descent & Cajuns (Louisiana)
 - strikes 1 in 3600 births
 - 100 times greater than incidence among non-Jews
 - non-functional enzyme fails to breakdown lipids in brain cells
 - fats collect in cells destroying their function
 - symptoms begin few months after birth
 - seizures, blindness & degeneration of muscle & mental performance
 - child usually dies before 5yo



AP Biology

Sickle cell anemia (recessive)

- Primarily Africans
 - strikes 1 out of 400 African Americans
 - high frequency
 - caused by substitution of a single amino acid in hemoglobin
 - when oxygen levels are low, sickle-cell hemoglobin crystallizes into long rods
 - deforms red blood cells into sickle shape
 - sickling creates pleiotropic effects = cascade of other symptoms



AP Biology

Sickle cell phenotype

- 2 alleles are codominant
 - both normal & mutant hemoglobins are synthesized in heterozygote (Aa)
 - 50% cells sickle; 50% cells normal
 - carriers usually healthy
 - sickle-cell disease triggered under blood oxygen stress
 - exercise

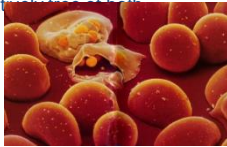


AP Biology

Heterozygote advantage

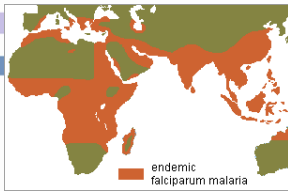


- Malaria
 - single-celled eukaryote parasite spends part of its life cycle in red blood cells
- In tropical Africa, where malaria is common:
 - **homozygous dominant** individuals die of malaria
 - **homozygous recessive** individuals die of sickle cell anemia
 - **heterozygote carriers** are relatively free of both
 - reproductive advantage
- High frequency of sickle cell allele in African Americans is vestige of African roots

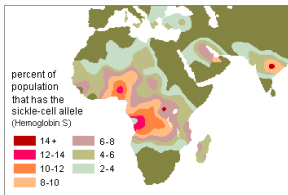


AP Biology

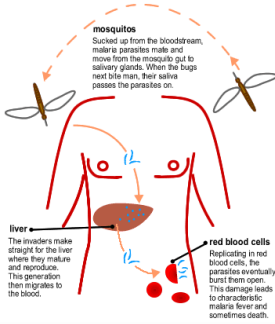
Prevalence of Malaria



Prevalence of Sickle Cell Anemia

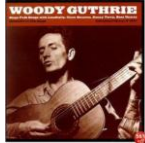
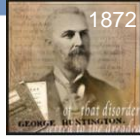


Malaria



Huntington's chorea (dominant)

- Dominant inheritance
 - repeated mutation on end of chromosome 4
 - mutation = CAG repeats
 - glutamine amino acid repeats in protein
 - one of 1st genes to be identified
- build up of "huntingtin" protein in brain causing cell death
 - memory loss
 - muscle tremors, jerky movements
 - "chorea"
 - starts at age 30-50
 - early death
 - 10-20 years after start



Testing...
Would you
want to
know?

AP Biology

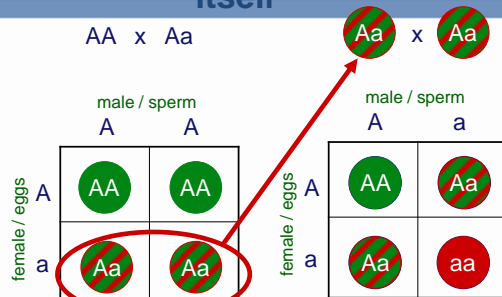
Genetics & culture

- Why do all cultures have a taboo against incest?
 - laws or cultural taboos forbidding marriages between close relatives are fairly universal
- Fairly unlikely that 2 unrelated carriers of same rare harmful recessive allele will meet & mate
 - but matings between close relatives increase risk
 - "consanguineous" (same blood) matings
 - individuals who share a recent common ancestor are more likely to carry same recessive alleles



AP Biology

A hidden disease reveals itself



- increase carriers in population
- hidden disease is revealed

AP Biology
