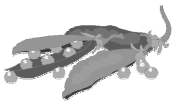
Psych 3102
Lecture 3

Mendelian Genetics

Gregor Mendel

- 1822 – 1884
- Augustinian monk
- paper read 1865-66
- genotype

- phenotype



Keys to Mendel's success
- good scientific method

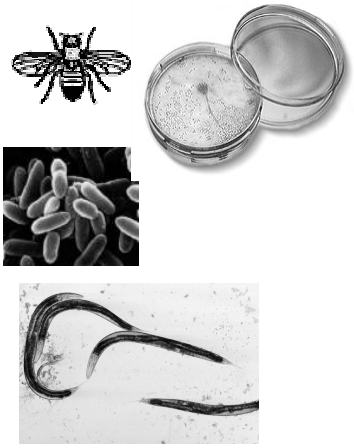
Good choice of organism

- pea plants
- Drosophila
- bacteria, molds
- Caenorhabiditis elegans

self-fertilization

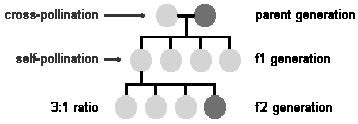
cross-fertilization

cloning



Vocabulary

- pure (true)- breeding
- hybrid
- monohybrid cross
- dihybrid cross

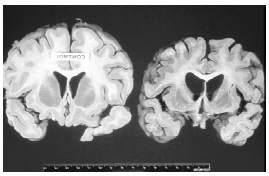


P F_1 F_2

Monohybrid cross

Examples:

Huntington disease (HD)



Phenylketonuria (PKU)

Both are single gene disorders that effect the nervous system

Huntington disease

- Phenotype (characteristics)

personality changes memory loss involuntary spasms
complete loss of motor control and intellectual functioning
late onset (~ 40 years)
lethal within 15-20 years

- Prevalence

1 in 20,000 live births (rare)

- Inheritance pattern

affected person always has one affected parent males and females affected
half of children in such a family are typically affected

- Let H = allele for HD
  h = normal allele

Cross between an affected person and an unaffected person:

P Hh x hh
affected normal

F_1 Punnet square:

Affected person always dies, allele would be expected to get selected out of human population. Why is it still present?

Phenylketonuria

Phenotype (characteristics)

mental retardation (post-natal brain damage)
affected person cannot metabolize phenylalanine

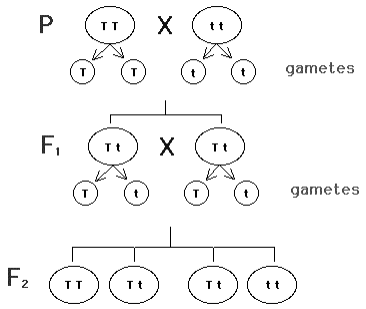
Prevalence

1 in 10,000 live births

Inheritance pattern

affected person can have 2 normal parents, males and females equally affected
1 in 4 children in such a family are typically affected

If T = normal allele
t = PKU allele



Outcome when one parent is homozygous normal and the other is a normal carrier?

Outcome when one parent is affected and one is a carrier?

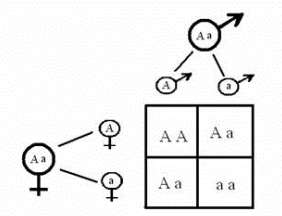
Why is PKU more common in consanguineous marriages?

1 in 50 people in the general population are carriers of the PKU allele.

Mendel's Laws of Heredity

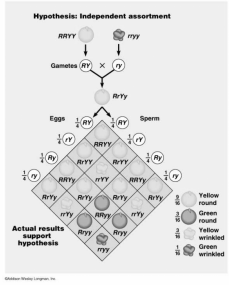
1. LAW OF SEGREGATION (MENDEL'S FIRST LAW)

2 alleles for each gene in each person
alleles separate (segregate) during reproduction
offspring receive 1 allele from each parent



2. LAW OF INDEPENDENT ASSORTMENT (MENDEL'S SECOND LAW)

alleles for different genes segregate independently



The second law is only true if the genes for the traits are on different chromosomes

- linkage
  - when genes for two traits are on the same chromosome

linkage analysis – used to detect linkage
- can be used to locate genes to chromosomes

dihybrid cross – used in linkage analysis for two single-gene traits
linked genes gives ratios that differ from the expected 9:3:3:1

Huntington disease

- linkage analysis with a large pedigree and markers used to locate gene to chromosome 4p in 1983
- finer mapping using markers near the suspected location pin-pointed the exact position of the gene in 1993
- genetic test now available
- huntingtin protein product being investigated
- trinucleotide repeat mutation

PKU

- located to chromosome 12 using its product, 1984
- blood test used to detect PKU at birth
- DNA test difficult due to # mutations
- strict diet prevents brain damage
- many different point mutations