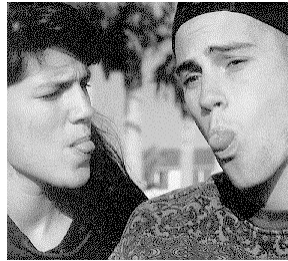


# Psych 3102

## Lecture 4

### Mendelian genetics in humans



## Problems

- no controlled mating
- unknown genetic background
- long generation time
- small family size
- no environmental control

To look for Mendelian inheritance patterns, employ the use of

large pedigrees – large family trees showing relationships and phenotypes

pedigree analysis – try to infer genotypes and hence inheritance patterns

- Symbols used in human pedigree analysis
- autosomal recessive traits
- autosomal dominant traits

- deleterious harmful

If the allele producing a deleterious trait is dominant, the individual is almost always heterozygous. Why?

Deleterious dominants survive in the population by exhibiting:

- variable expressivity people with the same genotype show varying phenotypic expressions
- low (incomplete) penetrance some people with a particular genotype do not show the expected phenotype

60% with Aa show expected phenotype (ie allele is penetrant in these people)

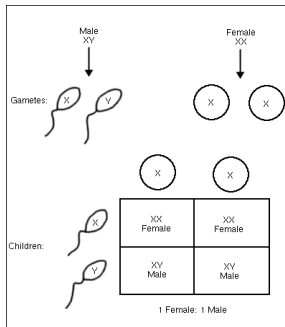
40% with Aa do not show expected phenotype (ie. allele is not penetrant in these people) Penetrance of this allele = 60% , ie it shows low or incomplete penetrance in the population

## Beyond Mendel

### - Extensions of Mendelian genetics

#### Sex Linkage

- genes for the trait are on the X or Y chromosome



- genes on male parent X never inherited by his sons
- genes inherited from mother's X will always be expressed in a son

# Example – Duchenne muscular dystrophy (DMD)

Inheritance: X-linked recessive

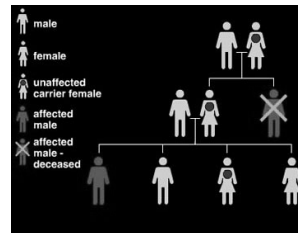
Prevalence: 1 in 3500 males

Phenotype: neuromuscular disorder

progressive wasting of muscles

death by age 20

neurons in brain also affected



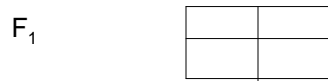
## Inheritance pattern for X-linked recessive alleles

- Example

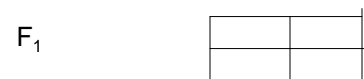
$X^D$  = normal allele on X chromosome

$X^d$  = Duchenne dystrophy allele

P normal mother x affected father



P carrier mother x normal father





## Y-linked inheritance holandric traits

1. Never shown in females
2. every male with the allele will express it

TDF (SRY) testis-determining factor

possibly – hairy ear syndrome