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

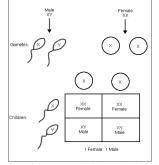
 varying phenotypic expressions
 low (incomplete) penetrance some people with a particular genotype do not show the expected phenotype

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

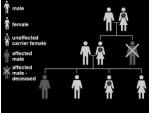


- genes on male parent X never inherited by his sons
- genes inherited from mother's X will always be expressed in a son

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 Xd = Duchenne dystrophy allele

P normal mother x affected father

 F_1

P carrier mother x normal father

 F_1

Inheritance pattern for X-linked dominant allele

- 1. affected male will have no normal daughters but no affected sons
- 2. heterozygous female transmits to 50% progeny of either sex
- 3. affected females are more common than affected males

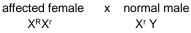
examples: webbing of toes
<u>Rett syndrome</u>

1 in 10,000 girls (lethal in boys) mental and physical disability after first year of life spontaneous mutation



X^{R} = Rett allele on X chromosome
X ^r = normal allele

Ρ







х



Reciprocal cross (reverse phenotypes of sexes) shows sex differences

Rett male

Р

 F_1

- (hypothetical case)

normal female

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