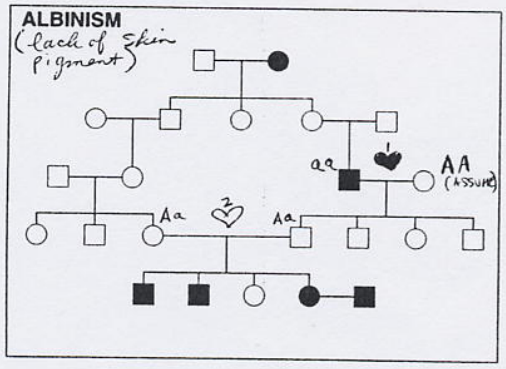
AUTOSOMAL RECESSIVE:

DR NEDWIDEK
SB3P/49
2/15/2008
2/2009
SL2 5/2009

Ex

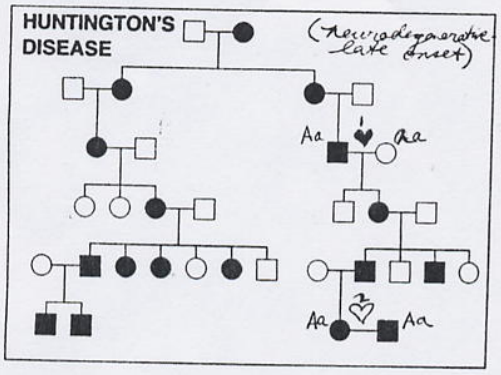


Characteristics of Autosomal Recessive Inheritance

1. An autosomal recessive phenotype, if it appears in more than one member of a kindred, typically is seen only in the sibship of the proband, not in parents, offspring, or other relatives.
2. For most autosomal recessive diseases, males and females are equally likely to be affected.
3. Parents of an affected child are asymptomatic carriers of mutant alleles.
4. The parents of the affected person may in some cases be consanguineous. This is especially likely if the gene responsible for the condition is rare in the population.
5. The recurrence risk for each sib of the proband is 1 in 4.

AUTOSOMAL DOMINANT:

Ex



Characteristics of Autosomal Dominant Inheritance

1. The phenotype usually appears in every generation, each affected person having an affected parent.

   Exceptions or apparent exceptions to this rule in clinical genetics: (1) cases originating by fresh mutation in a gamete of a phenotypically normal parent; (2) cases in which the disorder is not expressed (nonpenetrant) or is expressed only subtly in a person who has inherited the responsible gene.
2. Any child of an affected parent has a 50 percent risk of inheriting the trait.

   This is true for most families, in which the other parent is phenotypically normal. Because statistically each family member is the result of an "independent event," wide deviation from the expected 1:1 ratio may occur by chance in a single family.
3. Phenotypically normal family members do not transmit the phenotype to their children.

   Failure of penetrance or very subtle expression of a condition may lead to apparent exceptions to this rule.
4. Males and females are equally likely to transmit the phenotype, to children of either sex. In particular, male-to-male transmission can occur, and males can have unaffected daughters.
5. A significant proportion of isolated cases are due to new mutation. The less the fitness is, the greater is the proportion due to new mutation.

EXCERPTS FROM:
THOMPSON + THOMPSON, ELLIOTT