Inheritance III-cont.

Continue with analysis of two-gene studies:
- Probabilities
- Punnett Square
- Branch Diagram
- Introduction to Pedigree Analysis
tracing the inheritance patterns of human disorders

~10,000 known single-gene human traits
see OMIM (Online Mendelian Inheritance in Man)

most are rare* genetic diseases
allele causing disorder can be recessive OR dominant

* rare = incidence of disease is <1%

Blue box:
- Albinism
- Cystic fibrosis
- Sickle cell anemia
- Tay Sachs disease
- Thalassemia

Orange box:
- Huntington's Disease
- Achondroplasia
- Marfan Syndrome
- Brachydactyly
- Polydactyly
Standardized Pedigree Symbols
and Relationships

- **Male**
- **Female**
- **Sex unspecified**
- **Proband**
- **Affected**
- **Carrier (autosomal)**
- **Carrier (X-linked)**
- **Deceased**
- **Divorced**
- **Consanguinous mating**

- **Couple** (horizontal line connects mates)
- **Offspring** (vertical line connects parents with offspring)
- **Adopted in**
- **Adopted out**
- **Monozygotic twins**
- **Dizygotic twins**
- **Zygosity unknown**
Inheritance of albinism

Recessive traits
- unaffected parents of affected children are carriers
- unaffected parents may have affected children
- all the children of two affected individuals are affected
- often appears to “skip a generation”
Inheritance of albinism

- Who can be identified as a carrier?
- What is the genotype of the monozygotic twins?
- If the two-carrier couple have another child - what is the chance of that child being albino?
- For rare diseases, we can assume that an unrelated individual is not a carrier.

ex. the mom of the 3 carrier siblings is assumed to be homozygous dominant since Albinism is not common

Aa × Aa

1 : 2 : 1

AA: Aa: aa
rare recessive disorders often show up in consanguinous matings
**Dominant traits**

- appears in every generation (cannot skip!)
- at least one parent of an affected child is affected
- two affected parents may have unaffected children
- an affected parent will transmit the trait to 1/2 progeny
- no “carriers”
- can they be lethal?
some pedigrees can be inconclusive