Lecture 7 – Pedigree analysis
I. Pedigree
   A. what is it? - diagram of familial relationship and transmission of traits.
   B. how is it useful?

C. In general, genetic disorders are very rare.
D. 4 primary patterns of inheritance
   1. autosomal recessive
   2. autosomal dominant
   3. X-linked recessive
   4. X-linked dominant
   5. Y-linked
E. Commonly used symbols.

II. Patterns of inheritance
   A. autosomal

   1. Clues to pattern:
      a. Unaffected parents can have affected offspring.
      b. Both sexes affected equally.
      c. Tends to skip generations.
      d. Two affected parents produce all affected children.
e. One effected and one unaffected parent usually produce all unaffected.
f. Trait is more frequent among offspring of related parents.

2. Examples:
   a. albinism
   b. cystic fibrosis
   c. sickle cell anemia
   d. phenylketonuria
   e. methylmalonic acidemia

B. autosomal dominant

1. Clues to pattern:
   a. Trait tends to appear in every generation.
   b. Both sexes affected equally.
   c. Unaffected parents produce only unaffected offspring.
   d. Two effected parents can produce unaffected children.
   e. On average, half the children of an effected parent will be affected.

2. Examples:
   a. Huntington’s chorea
   b. pseudo-achondroplasia (dwarfism)
   c. brachydactyly

C. X-linked recessive
1. Clues to pattern:
   a. Effected males more common than females.
   b. Effected fathers transmit gene to daughters, not sons.
   c. None of sone of effected father are affected or carry trait (unless mother carries)
   d. On average, half the sons of woman whose father was affected will be affected.
   e. A daughter will be affected only if her father is affected and her mother is at least heterozygous.

2. Examples:
   a. hemophilia A
   b. color blindness
   c. Duchenne muscular dystrophy
   d. Lesch-Nyhan

D. X-linked dominant

1. Clues to pattern:
   a. Effected males never have effected sons (unless mom affected).
   b. All daugthers of effected males are affected.
   c. On average, half the sons and daughters of effected mothers are affected.

2. Example:
   a. hypophosphatemia

E. Y-linked (holandric)

1. Clues to pattern:
   a. Transmitted from fathers to all sons.
   b. No females exhibit trait.

2. Example:
   a. possibly hairy ears
III. Recognizing patterns of inheritance
   A. dominant vs recessive
      recessive          dominant

   B. autosomal recessive vs X-linked recessive
      autosomal recessive  X-linked recessive

   C. Autosomal dominant vs. X-linked dominant.
      If daughter of effected male normal, or son of effected male is affected (unless mom also affected), then it must be autosomal dominant.

   D. Examples:
      Eg. 1:

      Eg. 2:
Eg. 3:

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I

II
1 2 3 4 5

III
1 2 3 4 5

IV
1 2 3 4

V
1 2 3 4
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Eg. 4:

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I

II
1 2 3 4 5

III
1 2 3 4 5 6 7
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