<table>
<thead>
<tr>
<th>Trait</th>
<th>Phenotype</th>
<th>OMIM Number</th>
</tr>
</thead>
<tbody>
<tr>
<td>Achondroplasia</td>
<td>Dwarfism associated with defects in growth regions of long bones</td>
<td>100800</td>
</tr>
<tr>
<td>Brachydactyly</td>
<td>Malformed hands with shortened fingers</td>
<td>112500</td>
</tr>
<tr>
<td>Camptodactyly</td>
<td>Stiff, permanently bent little fingers</td>
<td>114200</td>
</tr>
<tr>
<td>Crouzon syndrome</td>
<td>Defective development of mid-face region, protruding eyes, hook nose</td>
<td>123500</td>
</tr>
<tr>
<td>Ehlers-Danlos syndrome</td>
<td>Connective tissue disorder, elastic skin, loose joints</td>
<td>130000</td>
</tr>
<tr>
<td>Familial hypercholesterolemia</td>
<td>Elevated levels of cholesterol; predisposes to plaque formation, cardiac disease; may be most prevalent genetic disease</td>
<td>144010</td>
</tr>
<tr>
<td>Adult polycystic kidney disease</td>
<td>Formation of cysts in kidneys; leads to hypertension, kidney failure</td>
<td>173900</td>
</tr>
<tr>
<td>Huntington disease</td>
<td>Progressive degeneration of nervous system; dementia; early death</td>
<td>143100</td>
</tr>
<tr>
<td>Hypercalcemia</td>
<td>Elevated levels of calcium in blood serum</td>
<td>143880</td>
</tr>
<tr>
<td>Marfan syndrome</td>
<td>Connective tissue defect; death by aortic rupture</td>
<td>154700</td>
</tr>
<tr>
<td>Nail-patella syndrome</td>
<td>Absence of nails, kneecaps</td>
<td>161200</td>
</tr>
<tr>
<td>Porphyria</td>
<td>Inability to metabolize porphyrins; episodes of mental derangement</td>
<td>176200</td>
</tr>
</tbody>
</table>

For each of these conditions, mutant allele is dominant over wild type allele.
Marfan Syndrome Is an Autosomal Dominant Trait

- Affects skeletal system, eyes, and cardiovascular system
- Characteristics – tall, thin arms and legs with long, thin fingers and toes
- Weakened aorta may enlarge and rupture

Fig. 4.13
Marfan Syndrome May Affect the Aorta

Gene is located on chromosome 15 = fibrillin

Fig. 4.14
Pedigree of a dominant trait

Huntington’s Rare dominant trait

Consanguineous mating
Pedigree - Woody Guthrie
Huntington’s disease-affected brains

Figure D-4: Effect of HD on the Basal Ganglia

The basal ganglia of the human brain, showing the impact of HD on brain structure in this region. Note especially that the brain of a person with HD has bigger openings due to the death of nerve cells in that region.

Source: Singer, Jonathan. Huntington's Disease. Online. Available at:
http://list-socrates.berkeley.edu/~jmp/HD.html
Nancy Wexler
The number of CAG repeats in huntingtin gene (htt) is directly correlated with probability of disease.
Pathology of Huntington’s disease

http://www.birf.info/art/people/media-stories/nd5.gif
Examples of Patterns of Inheritance

• Autosomal Recessive
• Autosomal Dominant
• X-Linked Dominant
• X-Linked Recessive
• Y-Linked Inheritance
• Mitochondrial Inheritance

Chapter 4  Human Heredity by Michael Cummings ©2006 Brooks/Cole-Thomson Learning
Transmission of X and Y Chromosomes

Fig. 4.16
X and Y Chromosomes

- Females carry two copies of the X chromosome and may be homozygous or heterozygous for a given trait.
- Males carry only one X chromosome and are **hemizygous** for all genes on the X chromosome.
- A male receives his Y chromosome from his father and his X from his mother.
X-linked Dominant Traits

- Affected males produce all affected daughters but no affected sons
- (Criss-cross inheritance)
- Heterozygous females will have unaffected and affected offspring; sons and daughters will be equally affected
Hypophosphatemia
Is an X-linked Dominant Trait

Fig. 4.17
X-Linked Recessive

- Hemizygous males and homozygous females are affected
- Males are more likely to be affected
- Affected males receive the allele from their mothers
- Daughters must receive the allele from both parents
Pedigree for X-Linked Recessive Trait

Fig. 4.18
Colorblindness

- Most common form of color blindness is red-green blindness
- 8% male population of US
- Red blindness – unable to see red as a distinct color
- Green blindness – cannot see green or other colors in the middle of the visual spectrum
- Rare form is an autosomal dominant trait on chromosome 7 - blue blindness
Ishihara plate test for colorblindness
Defects in the Cones Cause Color Blindness

Fig. 4.21
Red-green opsin genes

Representative X chromosomes
(each male has only one)

1  Normal vision
2  Normal vision
3  Normal vision
4  Severe red-green color blindness
5  
6  Moderately severe
7  Mild

= green-pigment gene
= red-pigment gene
* = mutation in red-pigment gene

http://home.comcast.net/~john.kimball1/BiologyPages/R/Red-greenGenes.gif
Muscular Dystrophy

- Progressive weakness and loss of muscle tissue
- Autosomal and X-linked forms
- Most common form is an X-linked recessive **Duchenne muscular dystrophy**
- 1/3500 males in U.S.
- Disease progresses rapidly
Duchenne Muscular Dystrophy

- Gene located near end of X chromosome
- Encodes for dystrophin
- Stabilizes cell membrane during the stress of muscle contraction
- Several mutations with variation in phenotype
- Characteristic movements

Fig 4.22
Hemophilia and History