Y-Linked Traits

- Only males have Y chromosomes
- Passed from father to sons
- All Y-linked traits are expressed
- Approximately three dozen Y-linked traits have been discovered
Pedigree for Y-Linked Trait

Fig. 4.24
# Y-Linked Genes

## Table 4.4  Some of the Genes Mapped to the Y Chromosome

<table>
<thead>
<tr>
<th>Gene</th>
<th>Product</th>
<th>OMIM Number</th>
</tr>
</thead>
<tbody>
<tr>
<td>ANT3 ADP/ATP translocase</td>
<td>Enzyme that moves ADP into, ATP out of mitochondria</td>
<td>403000</td>
</tr>
<tr>
<td>CSF2RA</td>
<td>Cell surface receptor for growth factor</td>
<td>425000</td>
</tr>
<tr>
<td>MIC2</td>
<td>Cell surface receptor</td>
<td>450000</td>
</tr>
<tr>
<td>TDF/SRY</td>
<td>Protein involved in early stages of testis differentiation</td>
<td>480000</td>
</tr>
<tr>
<td>H-Y antigen</td>
<td>Plasma membrane protein</td>
<td>426000</td>
</tr>
<tr>
<td>ZFY</td>
<td>DNA binding protein that may regulate gene expression</td>
<td>490000</td>
</tr>
</tbody>
</table>
Mitochondrial Inheritance

- Mitochondria are cytoplasmic organelles important in cellular respiration
- Have their own DNA
- Carry 37 genes
- Transmitted from mother to ALL of her offspring
- No recombination
- Males and females equally affected
- High mutation rate
Pedigree of Mitochondrial Inheritance

Fig. 4.25
# Mitochondrial Genes

<table>
<thead>
<tr>
<th>Trait</th>
<th>Phenotype</th>
<th>OMIM Number</th>
</tr>
</thead>
<tbody>
<tr>
<td>Kearns-Sayre syndrome</td>
<td>Short stature; retinal degeneration</td>
<td>530000</td>
</tr>
<tr>
<td>Leber optic atrophy</td>
<td>Loss of vision in center of visual field; adult onset</td>
<td>535000</td>
</tr>
<tr>
<td>MELAS syndrome</td>
<td>Episodes of vomiting, seizures, and stroke-like episodes</td>
<td>540000</td>
</tr>
<tr>
<td>MERRF syndrome</td>
<td>Deficiencies in the enzyme complexes associated with energy transfer</td>
<td>545000</td>
</tr>
<tr>
<td>Oncocytoma</td>
<td>Benign tumors of the kidney</td>
<td>553000</td>
</tr>
</tbody>
</table>
Complications in genetic analyses

Pleiotropv

One gene influences more than one phenotype/trait

Mouse *agouti* gene
## Mouse *agouti* gene

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>AA</strong></td>
<td>agouti</td>
</tr>
<tr>
<td></td>
<td><em>(dark gray)</em></td>
</tr>
<tr>
<td><strong>A^Y_A</strong></td>
<td>yellow</td>
</tr>
<tr>
<td></td>
<td><em>dominant coat color</em></td>
</tr>
<tr>
<td><strong>A^Y_A^Y</strong></td>
<td>lethal</td>
</tr>
<tr>
<td></td>
<td><em>recessive lethal</em></td>
</tr>
</tbody>
</table>
The Manx cat

tail viability
Pleiotropy: Single gene can cause more than one phenotype

Phenotype: red hair, pale skin, and freckles.

A variant of a gene that encodes a protein that controls balance of pigments in the skin (melanocortin-1-receptor)
Pleiotropy: One Gene—More Than One Phenotype

**Huntington’s**: progressive dementia onset around 40-50 years

- involuntary movements \[\text{Phenotype 1}\]
- death in about 5 years after onset of disease \[\text{Phenotype 2}\]
Mendel

Each gene determines a single trait

Extension

Pleiotropy: one gene influences more than one trait

Different ratios depending on dominance/recessive relation for each trait
Genetic heterogeneity: one phenotype: many genes

- Autosomal recessive heterogenetic traits:
  - Hearing Loss: 132 genes
  - Albinism: several different genes
  - Blood clotting disorders
Complications in genetic analyses

The same genotype does not always produce the same phenotype

• Phenotype depends on penetrance
• Phenotype depends on expressivity
• Phenotype can be affected by other modifier genes
• Environment can affect phenotype
Penetrance

Not all individuals of a given genotype show the phenotype

Penetrance = \( \frac{\text{# showing phenotype}}{\text{Total of that genotype}} \) %

Neurofibromatosis: Autosomal Dominant

- neurofibromas all over body
  Penetrance: 50 to 80%
Complications in genetic analyses

The same genotype does not always produce the same phenotype

• Phenotype depends on penetrance
• Phenotype depends on expressivity
• Phenotype can be affected by other modifier genes
• Environment can affect phenotype
Expressivity refers to the degree to which an individual expresses the trait

- The severity of phenotype can vary (extreme to mild) in individuals of the same genotype

Retinoblastoma: An example of variable expressivity
- 25% are unaffected

- Among the 75% affected individuals, some are affected only in one eye while, others are affected in both eyes

- Affecting both penetrance and expressivity
Complications in genetic analyses

The same genotype does not always produce the same phenotype

• Phenotype depends on penetrance
• Phenotype depends on expressivity
• Phenotype can be affected by other modifier genes
• Environment can affect phenotype
Phenotype can be affected by other modifier genes

• Modifier genes alter the phenotype produced by other genes.

• Modifier genes by themselves may have little or no effect on the phenotype.

BbDd cross
Complications in genetic analyses

The same genotype does not always produce the same phenotype

• Phenotype depends on penetrance
• Phenotype depends on expressivity
• Phenotype can be affected by other modifier genes
• Environment can affect phenotype
Phenylketonuria (PKU)

Caused by a mutation in an enzyme:

**Phenylalanine Hydroxylase**

- **Phenylalanine** accumulates
- **Tyrosine** reduced
- **Melanin** reduced
- **Adrenaline** reduced
- **Phenylpyruvic acid**

Affects the Central Nervous System
Phenylketoneura (PKU)

• 1/12,000 live births

• Affected individual is retarded, has very fair skin and blue eyes (melanin reduction), also has less adrenaline.

• In US every newborn is screened for PKU

• Affected individual is put on a diet limited in phenylalanine

• Allows nearly normal development

• Environmental change - restricted diet can result in a profound change in phenotype