Patterns of Inheritance
Part 1

How are Traits passed on from Parents to Children?

Monogenic & Polygenic

- ABO blood group
- Rhesus hemolytic disease
- Hirschprung disease
- Adult stature

Mendelian (monogenic)  
polygenic
Patterns of Inheritance

- **Autosomal**
  - Dominant
  - Recessive

- **X-Linked**
  - Dominant
  - Recessive

- **Y-Linked**
Autosomal Dominant Inheritance

Specific Features

- Affect both males and females in equal proportions
- Transmitted from one generation to the next
- All forms of transmission between the sexes

- Affected individual usually have at least one affected parent
- Two affected parents having unaffected offspring

Autosomal Dominant Pedigree
**Pleiotropy**

A single gene that may give rise to two or more apparently unrelated effects. In tuberous sclerosis, learning difficulties, epilepsy, a facial rash known as adenoma sebaceum and subungual fibromas.


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**Variable Expressivity**

The clinical features variation from person to person. This variation can be seen even in the same family.

Image from: Atlas Of Genetic Diagnosis and Counseling, 2006 Humana Press Inc.

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**Pedigree Shows Variable Expressivity**

 Heralded traits:
- hearing loss
- differently colored eyes
- white forelock
- premature gray

Pedigree chart showing genetic inheritance patterns.

Figure 6.14 Human Molecular Genetics, 4th ed. of the 4th Library

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Reduced Penetrance

- Individuals heterozygous for gene mutations of a certain autosomal dominant disorders with no abnormal clinical features
- Probably results from a combination of genetic and environmental factors
- Need to be taken into account when interpret family history information for autosomal dominant disorders

Pedigree Shows Reduced Penetrance

New Mutations

- The sudden appearance of a condition usually is a result of new mutation.
- In less dramatic conditions other explanations for the 'sudden' appearance of a disorder must be considered.
- New dominant mutations, in certain instances, have been associated with an increased age of the father
New Mutation

Homozygosity for AD Traits

- In some instances, affected individuals appear either to be more severely affected, or to have an earlier age of onset.
- The heterozygote with an intermediate phenotype is consistent with a haploinsufficiency loss-of-function mutation.
- Some dominantly inherited disorders, homozygous individuals are not more severely affected than heterozygotes.

Autosomal Recessive Inheritance
Autosomal Recessive Inheritance Specific Features

- Affects males and females in equal proportions.
- Usually affects only individuals in one generation.
- Consanguinity in the parents provides further support.
- Affected individual usually have normal parent.

Autosomal Recessive Pedigree

Pseudodominance
Locus Heterogeneity

- A disorder inherited in the same manner due to mutations in more than one gene
- For example hearing impairment/deafness
- Disorders with the same phenotype from different genetic loci are known as genocopies

Double heterozygotes

Mutational Heterogeneity

- Different mutations in the same gene as being responsible
- Individuals who have two different mutations at the same locus and are known as compound heterozygotes
- Most individuals affected with an autosomal recessive disorder are probably compound heterozygotes rather than true homozygotes.