

Genetics: Mendelian Genetics (2)

Patterns of Inheritance

Outline

- 6. Extending Mendelian Genetics
 - a. Incomplete Dominance
 - b. Codominance
 - c. Polygenic Inheritance
 - d. Environmental Impact
- 7. Mendelian Inheritance in Humans
- 8. Key Terms
- 9. Conclusions

Incomplete dominance

Incomplete dominance – heterozygous phenotype is intermediate between two parents

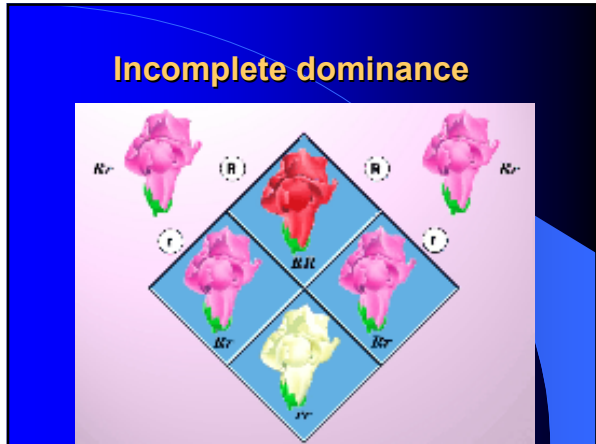
for example, snapdragons:

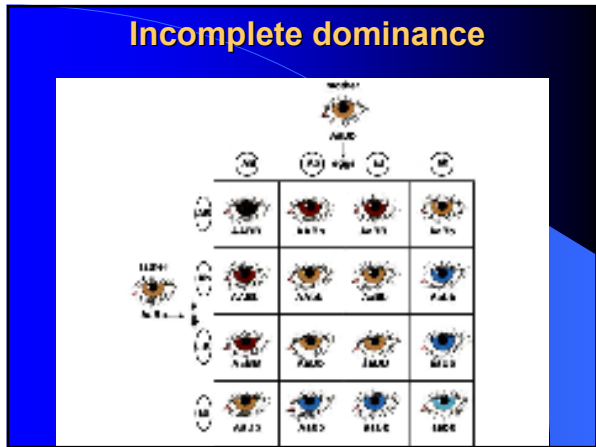
P: Red x White ($RR \times R'R'$)

F1: Pink (RR')

F2: $\frac{1}{4}$ Red(RR), $\frac{2}{4}$ Pink(RR'),

$\frac{1}{4}$ White($R'R'$)





Codominance

PHENOTYPE (BLOOD GROUP)	GENOTYPES	ANTIBODIES PRESENT IN BLOOD SERUM	REACTS (CLUMPS) WHEN RED BLOOD CELLS FROM GROUPS BELOW ARE ADDED TO SERUM FROM GROUPS AT LEFT?			
			O	A	B	AB
O	ii	Anti-A Anti-B	No	Yes	Yes	Yes
A	I ^A I ^A or I ^A i	Anti-B	No	No	Yes	Yes
B	I ^B I ^B or I ^B i	Anti-A	No	Yes	No	Yes
AB	I ^A I ^B	—	No	No	No	No

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Blood Typing

Human Blood types





The gene located on chromosome 9

Three alleles I^A , I^B , i (4 types: A, B, AB, O)

Based on markers on red blood cells

[Blood Type]

1. A ($I^A I^A$, $I^A i$) anti B antibodies
2. B ($I^B I^B$, $I^B i$) anti A antibodies
3. AB ($I^A I^B$) none
4. O (ii) anti A and anti B antibodies

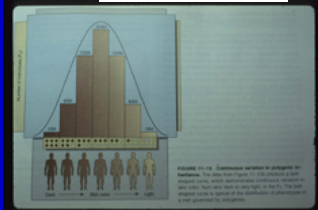
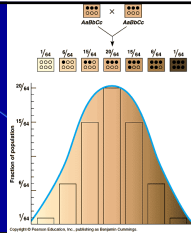
Genotype	Blood Type and Frequency in U.S.	Red Blood Cells	Cell Surface Protein	Cell Receptor Trc
$I^A I^A$ $I^A i$	A 40%	 A glycoprotein	A D	A AD
$I^B I^B$ $I^B i$	B 10%	 B glycoprotein	B D	B BD
$I^A I^B$ (Mutual receptor)	AB 4%	 Both A and B glycoproteins	A B D	AB
ii (Universal donor)	O 46%	 Neither A nor B glycoprotein	D	A B AB D

Polygenic inheritance

Polygenic inheritance – when alleles at more than one locus contribute to the same trait

for example:

- (1) skin color
- (2) human height



Polygenic inheritance



Environmental Impact





Mendelian Inheritance in Humans

1. Pedigree analysis
2. Recessive inherited diseases
3. Dominant inherited diseases

Pedigree analysis

First generation (grandparents)

Second generation (parents of a patient and sibling)

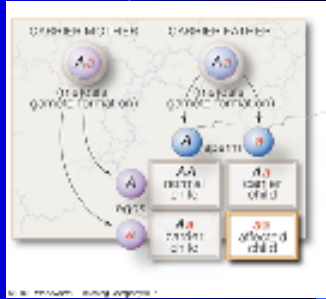
Third generation (patients)

10. Pedigree tracing is essential for genetic counseling

11. Pedigree tracing is necessary for affected siblings

Patterns of Autosomal Recessive Inheritance

Both Parents are carriers



Albinism

- Caused by autosomal recessive allele (aa)
- Absence of pigment
- Tyrosine (not a pigment)
 - > Melanin (pigment)
 - ↑
 - Tyrosinase



Sickle cell anemia

Sickle cell anemia is a recessive disease; one amino acid changes in hemoglobin molecule; Sickle-cell hemoglobin molecules tend to cluster together and block the capillary vessels.



normal SS, disease ss, carriers Ss.
In Africa, 45% of certain population have the genotype Ss. Because the heterozygotes (Ss) are resistant to the disease Malaria (a deadly disease).

Cystic Fibrosis

4% whites are carriers – the most common lethal genetic diseases in the US. Most children with cystic fibrosis die before five years old. Now may live to over 30s.

The gene can hide in the population for long time.

0.1% X 0.1% → it is still there

Recessive disease

Parents: Normal heterozygous for cystic fibrosis

P: Cc x Cc

[Punnett Square] ♂\♀

	C	c
C	CC	Cc
c	Cc	cc

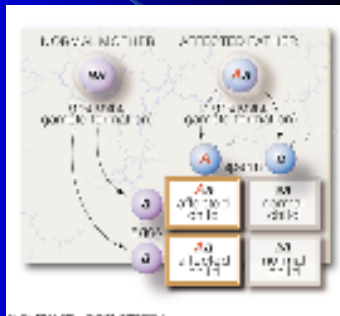
25% normal, 50% carriers, 25% with the disease

Patterns of Autosomal Dominant Inheritance

1. Achondroplasia
1/10,000

2. Huntington disorder
no symptoms until the carrier is about 35 to 45 years old

disease gene located on chromosome # 4



Patterns of Autosomal Dominant Inheritance

One type of dwarfism in humans is caused by a single dominant gene; the condition is called Achondroplastic dwarfism. Dwarf individuals are heterozygous, while persons who are homozygous recessive are of normal stature. The homozygous dominant individuals all die before birth. (20% family history, and 80% new mutations)
If two achondroplastic dwarfs marry and have a family, what fraction of their living offspring will also be achondroplastic dwarfs?

Dwarf Aa Normal aa AA die P: Aa X Aa

F1 genotypes: AA(die)Aa Aa aa 2/3 to be dwarf

Key Terms

Character	Law of segregation
Trait	Law of independent assortment
True-breeding	Incomplete dominance
Hybridization	Codominance
Monohybrid cross	Multiple alleles
Dihybrid cross	Polygenic inheritance
Alleles	Carrier
Dominant allele	Cystic fibrosis
Recessive allele	Sickle-cell disease
Homozygous	Achondroplasia
Heterozygous	Huntington disorder
Phenotype	
Genotype	

In Conclusion

1. *A gene is a unit of information about a heritable trait*
2. *Mendel provided evidence of dominant and recessive genes*
3. *Monohybrid crosses are crosses between two individuals that are homozygous for different versions of a trait*
4. *Crosses from F_1 result in F_2 offspring with phenotypes having a 3:1 ratio*
5. *Dihybrid crosses result in 9:3:3:1 phenotypic ratio*

In Conclusion

6. *Theory of Independent Assortment states that gene pairs independently sort out into different gametes regardless of other gene pairs of other chromosomes*
7. *Gene dominance can be in different level from complete dominance to incomplete dominance to co-dominance*
