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 x R'R')
F1: Pink (RR')
F2: 1/4 Red(RR), 2/4 Pink(RR'), 1/4 White(R'R')
Incomplete dominance

Codominance

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

<table>
<thead>
<tr>
<th>Blood Type</th>
<th>Phenotype</th>
<th>Antibodies</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>I^A I^A, I^A i</td>
<td>anti B antibodies</td>
</tr>
<tr>
<td>B</td>
<td>I^B I^B, I^B i</td>
<td>anti A antibodies</td>
</tr>
<tr>
<td>AB</td>
<td>I^A I^B</td>
<td>none</td>
</tr>
<tr>
<td>O</td>
<td>ii</td>
<td>anti A and anti B antibodies</td>
</tr>
</tbody>
</table>
Polygenic inheritance

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

(1) skin color
(2) human height
Environmental Impact

Mendelian Inheritance in Humans

1. Pedigree analysis
2. Recessive inherited diseases
3. Dominant inherited diseases
4. New tools for genetic testing

Pedigree analysis

First generation (grandparents)
Second generation (parents plus aunts and uncles)
Third generation (two sisters)

(a) Pedigree tracing a dominant trait (widow's peak)
(b) Pedigree tracing a recessive trait (attached earlobe)
Pedigree analysis

Patterns of Autosomal Recessive Inheritance

Both Parents are carriers

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).
Sickle cell anemia

Pleiotropic effects
One gene has multiple phenotypic effects

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 20s.
The gene can hide in the population for long time.
0.1% X 0.1% \( \rightarrow \) it is still there
Recessive disease
Parents: Normal heterozygous for cystic fibrosis
\[ P: Cc \times Cc \]

\[ \begin{array}{c|c|c}
\text{Genotype} & \text{Normal} & \text{Carriers} \\
\hline
\text{Cc} & 25\% & 50\% \\
\text{cc} & 25\% & 0\%
\end{array} \]

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  Polygenic inheritance
Dihybrid cross  Pleiotropy
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 F1 result in F2 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 levels from complete dominance to incomplete dominance to co-dominance.