The relationship between genotype and phenotype is rarely simple

- In the 20th century, geneticists have extended Mendelian principles not only to diverse organisms, but also to ***patterns of inheritance*** more complex than Mendel described.
- In fact, Mendel had the good fortune to choose a system that was relatively simple (genetically speaking).
  - Each character is controlled by a single gene.
  - Each gene has only two alleles, one of which is completely dominant to the other.
- The heterozygous F_1 offspring of Mendel’s crosses always looked like one of the parental varieties because one allele was dominant to the other.
- However, some alleles show ***incomplete dominance*** where heterozygotes show a distinct intermediate phenotype, not seen in homozygotes.
  - This is not blended inheritance because the traits are separable (particulate) as seen in further crosses.
  - Offspring of a cross between heterozygotes will show three phenotypes: both parentals and the heterozygote.
  - The phenotypic and genotypic ratios are identical, 1:2:1.
- A clear example of incomplete dominance is seen in flower color of snapdragons.
- A cross between a white-flowered plant and a red-flowered plant will produce all pink F_1 offspring.
- Self-pollination of the F_1 offspring produces 25% white, 25% red, and 50% pink offspring.
- ***Complete*** and ***incomplete*** dominance are part of a spectrum of relationships among alleles.
- At the other extreme from complete dominance is ***codominance*** in which two alleles affect the phenotype in separate, distinguishable ways.
  - For example, the M, N, and MN blood groups of humans are due to the presence of two specific molecules on the surface of red blood cells.
  - People of group M (genotype MM) have one type of molecule on their red blood cells, people of group N (genotype NN) have the other type, and people of group MN (genotype MN) have both molecules present.

- Because an allele is dominant does not necessarily mean that it is more common in a population than the recessive allele.
  - For example, polydactyly, in which individuals are born with extra fingers or toes, is due to an allele dominant to the recessive allele for five digits per appendage.
  - However, the recessive allele is far more prevalent than the dominant allele in the population.
    - 399 individuals out of 400 have five digits per appendage.
- Dominance/recessiveness relationships have three important points.

  1. They range from complete dominance, though various degrees of incomplete dominance, to codominance.

  2. They reflect the mechanisms by which specific alleles are expressed in the phenotype and do not involve the ability of one allele to subdue another at the level of DNA.

  3. They do not determine or correlate with the relative abundance of alleles in a population.
- Most genes have more than two alleles in a population.
- The ABO blood groups in humans are determined by three alleles, I^A, I^B, and i.
  - Both the I^A and I^B alleles are dominant to the i allele
  - The I^A and I^B alleles are codominant to each other.
- Because each individual carries two alleles, there are six possible genotypes and four possible blood types.
  - Individuals that are $I^A I^A$ or I^A i are type A and place type A oligosaccharides on the surface of their red blood cells.
  - Individuals that are $I^B I^B$ or I^B i are type B and place type B oligosaccharides on the surface of their red blood cells.
  - Individuals that are $I^A I^B$ are type AB and place both type A and type B oligosaccharides on the surface of their red blood cells.
  - Individuals that are ii are type O and place neither oligosaccharide on the surface of their red blood cells.
- Matching compatible blood groups is critical for blood transfusions because a person produces antibodies against foreign blood factors.
  - If the donor's blood has an A or B oligosaccharide that is foreign to the recipient, antibodies in the recipient's blood will bind to the foreign molecules, cause the donated blood cells to clump together, and can kill the recipient.

- The genes that we have covered so far affect only one phenotypic character.
- However, most genes are **pleiotropic**, affecting more than one phenotypic character.

Pleiotropy – the ability of a gene to affect an organism in many ways

  - For example, the wide-ranging symptoms of sickle-cell disease are due to a single gene.

- In **epistasis**, a gene at one locus alters the phenotypic expression of a gene at a second locus.
  - For example, in mice and many other mammals, coat color depends on two genes.
  - One, the epistatic gene, determines whether pigment will be deposited in hair or not.
    - Presence (C) is dominant to absence (c).
  - The second determines whether the pigment to be deposited is black (B) or brown (b).
    - The black allele is dominant to the brown allele.
  - An individual that is cc has a white (albino) coat regardless of the genotype of the second gene.
- A cross between two black mice that are heterozygous (BbCc) will follow the law of independent assortment.
- However, unlike the 9:3:3:1 offspring ratio of an normal Mendelian experiment, the ratio is nine black, three brown, and four white.
- Some characters do not fit the either-or basis that Mendel studied.
- **Quantitative characters** vary in a population along a continuum
- These are usually due to **polygenic inheritance,** the additive effects of two or more genes on a single phenotypic character.
  - For example, skin color in humans is controlled by at least three different genes.
  - Imagine that each gene has two alleles, one light and one dark, that demonstrate incomplete dominance.
  - An AABBCC individual is dark and aabbcc is light.
- A cross between two AaBbCc individuals (intermediate skin shade) would produce offspring covering a wide range of shades.
  - Individuals with intermediate skin shades would be the most likely offspring, but very light and very dark individuals are possible as well.
  - The range of phenotypes forms a normal distribution.

- Phenotype depends on environment and genes.
  - A single tree has leaves that vary in size, shape, and greenness, depending on exposure to wind and sun.
  - For humans, nutrition influences height, exercise alters build, sun-tanning darkens the skin, and experience improves performance on intelligence tests.
  - Even identical twins, genetic equals, accumulate phenotypic differences as a result of their unique experiences.
- The relative importance of genes and the environment in influencing human characteristics is a very old and hotly contested debate.
- The product of a genotype is generally not a rigidly defined phenotype, but a range of phenotypic possibilities, the **norm of reaction**, that are determined by the environment.
  - In some cases the norm of reaction has no breadth (for example, blood type).
- Norms of reactions are broadest for polygenic characters.
  - For these **multifactorial characters**, environment contributes to their quantitative nature.