Supplementary Material to
Extensions to Mendelian Genetics

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BIO 184
Dr. Tom Peavy

Incomplete Penetrance

• In some instances, a dominant allele is not expressed in a heterozygote individual
• Example = Polydactyly
  – Autosomal dominant trait
  – Affected individuals have additional fingers and/or toes
  – A single copy of the polydactyly allele is usually sufficient to cause this condition
  – In some cases, however, individuals carry the dominant allele but do not exhibit the trait
Inherited the polydactyly allele from his mother and passed it on to a daughter and son

Does not exhibit the trait himself even though he is a heterozygote

**Incomplete Penetrance**

- The term indicates that a dominant allele does not always “penetrate” into the phenotype of the individual

- The measure of penetrance is described at the population level
  - If 60% of heterozygotes carrying a dominant allele exhibit the trait allele, the trait is 60% penetrant

- Note:
  - In any particular individual, the trait is either penetrant or not
Expressivity

- Expressivity is the degree to which a trait is expressed
- In the case of polydactyly, the number of digits can vary
  - A person with several extra digits has high expressivity of this trait
  - A person with a single extra digit has low expressivity

- The molecular explanation of expressivity and incomplete penetrance may not always be understood
- In most cases, the range of phenotypes is thought to be due to influences of the
  - Environment
    - and/or
  - Other genes
Environmental Influence

• Example = Phenylketonuria
  – Autosomal recessive disorder in humans
  – Caused by a defect in the gene that encodes the enzyme phenylalanine hydroxylase
    • Converts phenylalanine to tyrosine
  – Affected individuals cannot metabolize phenylalanine
    • Phenylalanine will thus accumulate
    • It ultimately causes a number of detrimental effects
      – Mental retardation, for example

• Phenylketonuria
  – Newborns are now routinely screened for PKU
  – Individuals with the disease are put on a strict dietary regimen
    • Their diet is essentially phenylalanine-free
  – These individuals tend to develop normally
  – Thus the PKU test prevents a great deal of human suffering
    • Furthermore, it is cost-effective
• Gene interactions occur when two or more different genes influence the outcome of a single trait

• Indeed, morphological traits such as height weight and pigmentation are affected by many different genes in combination with environmental factors

• The term EPISTASIS describes the situation in which a gene can mask the phenotypic effects of another gene

• Epistatic interactions often arise because two (or more) different proteins participate in a common cellular function
  – For example, an enzymatic pathway

• If an individual is homozygous for either recessive allele
  – It will not make any functional enzyme C or enzyme P
  – Therefore, the flowers remain white