Penetrance and Expressivity

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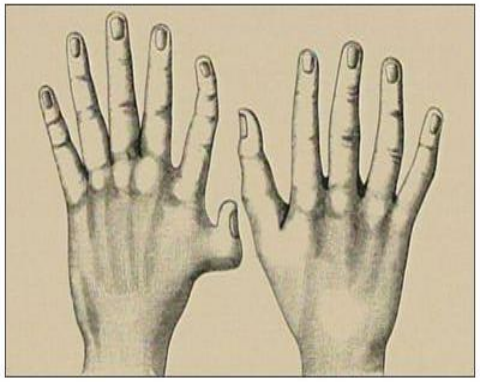
Penetrance

- The proportion of individual in population expressing in phenotype corresponding to genotype is called penetrance
- If all individual in population express the phenotypes corresponding to the genotypes then called Complete penetrance.
- If sum of the individual in the population failed to express a phenotype corresponding to genotype then it is called incomplete penetrance.
- Penetrance refers to the probability of a gene or trait being expressed. In some cases, despite the presence of a dominant allele, a phenotype may not be present. One example of this is polydactyly in humans (extra fingers and/or toes).

- A genetic condition to future generations. A dominant allele produces polydactyly in humans but not all humans with the allele display the extra digits. “Complete” penetrance means the gene or genes for a trait are expressed in all the population who have the genes. “Incomplete” or ‘reduced’ penetrance means the genetic trait is expressed in only part of the population. The penetrance of expression may also change in different age groups of a population.
- Reduced penetrance probably results from a combination of genetic, environmental, and lifestyle factors, many of which are unknown. This phenomenon can make it challenging for genetics professionals to interpret a person’s family medical history and predict the risk of passing

Polydactyly

- Genetically inherited diseases like Ellis-van Creveld are more concentrated among the Amish, because they marry within their own community.
- **This prevents new genetic variation from entering the population.** Children are therefore more likely to inherit 2 copies of the particular recessive genes that lead to genetic disease.



Expressivity

- Penetrance is quantified and have values ranging from (0-100%) the degree to which a genotype is expressed in different region of individual is called Expressivity.
- If all region of the body phenotype corresponding to genotype then it is called complete expressivity.
- If some region of body failed to show phenotype corresponding to the genotype then it is called incomplete expressivity. It can’t be quantified.
- Even though polydactyly is an autosomal dominant , most of the heterozygote remain pentdactyly five digits in each harm
- when individual express polydactyly it often limited to hand but not legs indicating incomplete expressivity.

- Expressivity on the other hand refers to variation in phenotypic expression when an allele is penetrant. Back to the polydactyly example, an extra digit may occur on one or more appendages. The digit can be full size or just a stub. Hence, this allele has reduced penetrance as well as variable expressivity.
- Variable expressivity refers to the range of signs and symptoms that can occur in different people with the same genetic condition. As with reduced penetrance, variable expressivity is probably caused by a combination of genetic, environmental, and lifestyle factors, most of which have not been identified. If a genetic condition has highly variable signs and symptoms, it may be challenging to diagnose.

Penetrance and Expressivity

- **Expressivity** refers to the severity or extent

A phenotype is variably expressive if symptoms vary in intensity among different people

EX: one person with polydactyly may have an extra digit on both hands and a foot, but, another may have just one extra fingertip.

Polydactyly is both *incompletely penetrant* and *variable expressive*.

Phenocopy

- By environmental change that inhibited for few generation as genetically inherited trait is called Phenocopy.
- Depending upon the extent to which the environment influences the genotype, the changes in the phenotype may be subtle or dramatic. Sometimes the phenotype becomes altered by the environment in such a way that the new phenotype resembles another phenotype produced by known genes. The induced phenotype is not inherited and is called a phenocopy.
- Phenotype doesn’t involve in change in the genotype and therefore it can’t be permanently inherited similarly to that genocopy .
- The first ever phenocopy studied was a decrease a size of Drosphila when they grow in on elevated temperature

In rabbits, the *Himalayan* allele is temperature dependent.

Reared at 20°C or less

Reared at temperatures above 30°C

Figure 5-20
Genetics: A Conceptual Approach, Third Edition

- The recessive gene vg/vg which produces vestigeal wings in Drosophila is also influenced by temperature. At 32°F the wings are feebly developed and extend very little from the body At 40°F the wings are better developed and have some venation. At 88°F wings are well developed with conspicuous venation.

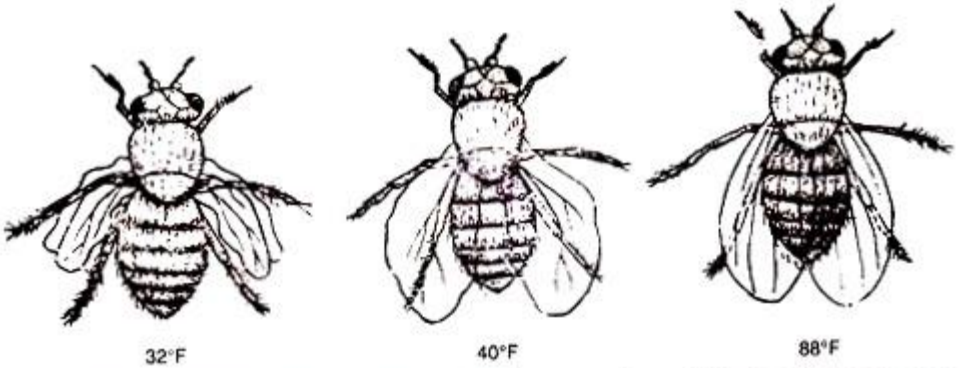


Fig. 3.1 Influence of temperature on the expression of the gene for vestigial wings (*vg*) in *Drosophila*.

EXAMPLE FOR ALL

- Diabetes mellitus is a heritable human trait associated with reduced amounts of the hormone insulin that is secreted by the pancreas. In the presence of insulin glucose is absorbed by the cell membranes. When the hormone is not produced in sufficient quantity, the unabsorbed glucose passes into the blood and urine. The exact mode of inheritance of diabetes is not properly understood.
- There are different types of diabetes arising from different causes; it therefore seems likely that there are several gene pairs controlling the trait. On the other hand the study of a pair of genetically identical twins, one of whom had diabetes the other not, indicates that the condition is due to a recessive gene with low penetrance.
- If proper doses of insulin are administered to a diabetic person, he reverts to the normal phenotype. In other words, control of diabetes produces a phenocopy of the normal individual. There are many other examples in human beings where, by giving drugs, the mutant genotype produces a phenocopy of the normal phenotype.