Chapter 8
Polygenic Inheritance and Pleiotropy

NCEA Level 3 Biology material is covered in this chapter for Achievement Standard 3.3 'Describe gene expression' by:

- Discussing biological concepts and processes relating to gene expression, through gene-gene interactions involving polygenes and pleiotropy, and gene-environment interactions.

Types of Variation

Discontinuous variation

This involves characters that are of the 'either-or' kind – eg blood groups, tongue-rolling ability, the ability to taste phenylthiocarbamide (PTC), or to distinguish visually between red and green. Mendel used discontinuously varying characters in his breeding experiments.

Continuous variation

In this kind of variation there is a smooth series of intermediates between two extremes, so there are no distinct groups. Human examples are height, skin colour, blood clotting time, blood pressure, and thickness of fat under the skin. Whereas with discontinuously varying characters organisms are *counted*, with continuous variation they are *measured*. When a large number of measurements have been obtained, the data can be plotted in the form of a histogram, with the number of individuals plotted against size of the measurement.

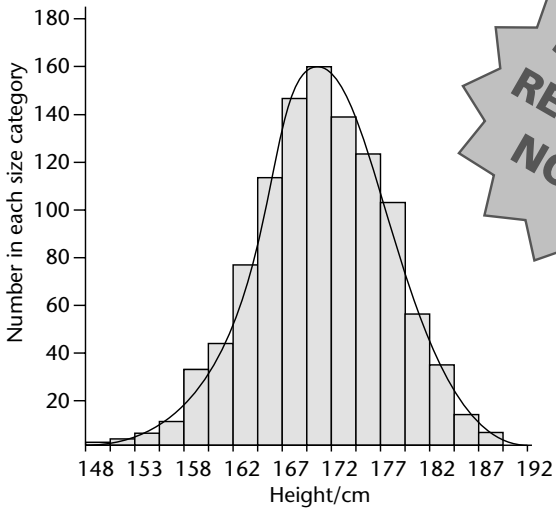


Fig. 8.1: *Variation in height amongst 1 083 adult men.*

Characteristics such as those mentioned above are controlled by many genes – called **polygenic inheritance**. Each individual gene has a small effect on the phenotype, and changing one allele for another at a particular locus for one of the genes has an even smaller effect. Genes in a polygenic system are inherited according to Mendelian principles.

Inheritance and Polygenes

Polygenes resulting in continuous variation can be explained by conventional genetics if we assume that:

- There are many loci involved, giving scope for a large number of genotypes. The addition of many individually small gene effects would give many phenotypes differing by small degrees.
- Many, perhaps most, continuously varying characters are sensitive to environmental variations (eg skin colour, body fat, blood pressure). Since no two organisms have identical environments, even individuals with the same genotype will show phenotypic variation.

To see how these two factors could act to produce a smooth distribution of phenotypes, suppose that the intensity of human skin pigmentation is under the influence of two pairs of alleles, and the following assumptions are made:

- Each of the two loci exists as two alleles *A/a* and *B/b* and there is no dominance, each allele of each gene is incompletely dominant to the other alleles.
- Each 'upper case' allele contributes one unit to the darkness of skin pigmentation, and each 'lower case' allele contributes none.
- Both loci have the same effect and the individual gene effects add together, so that in the same environment *AAbb* gives the same pigmentation as *aaBB* or *AaBb*.

The expected results of matings between *AaBb* × *AaBb* people could produce five shadings of skin colour:

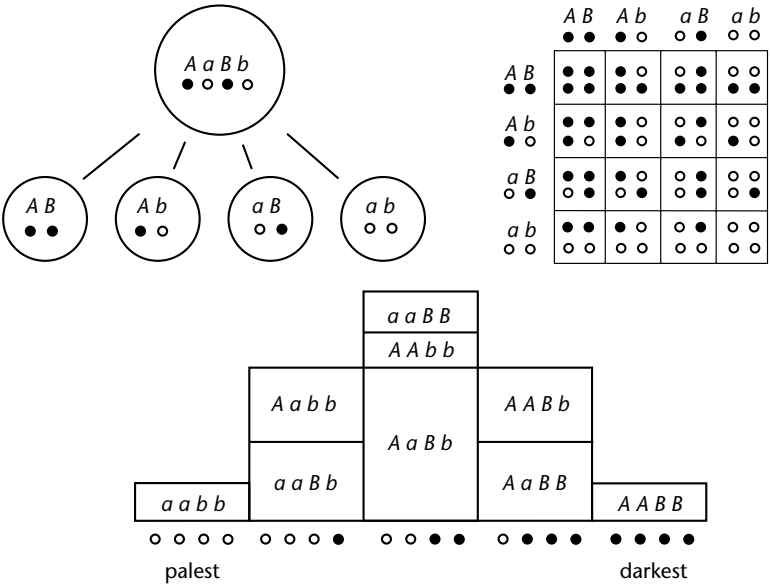


Fig. 8.2: *Incomplete dominance at two loci for skin colour.*

The situation is probably more complicated than that outlined, with as many as five gene loci probably being involved in skin colour. The more loci (and therefore alleles) involved, the greater the number of phenotypic classes. With no dominance there would be three genotypes

for each locus, giving a total of $3^5 = 243$ possible genotypes. However, many of these would have similar phenotypes; for instance there would be five genotypes with only one 'upper case' allele (*Aabbccddee, aaBbccddee, aabbCcddee, aabbccDdee* and *aabbccddEe*). Altogether there would be eleven phenotypes:

10 'upper case' alleles (1 genotype),
9 'upper case' alleles and 1 'lower case' allele (5 genotypes),
8 'upper case' and 2 'lower case' alleles,
7 'upper case' and 3 'lower case' alleles,

and so on, the eleventh phenotype as a result of 10 'lower case' alleles (one genotype). Moreover, there are environmental factors to be taken into account. No two people are exposed to identical amounts of UV, and exposure to UV determines suntan depth.

Worked Example

In a cross between a homozygous red-flowered plant and a homozygous white-flowered variety, approximately $\frac{1}{256}$ of the F_2 were white. Assuming that there are two alleles at each locus, how many gene loci appear to be involved in the production of pigment in this species?

Solution

If one locus were involved, then $\frac{1}{4}$ of the F_2 would be white. If there were two loci, $\frac{1}{16}$ would be white. Three loci would give $\frac{1}{64}$ white, and four loci would give $\frac{1}{256}$. Hence there are probably four loci involved.

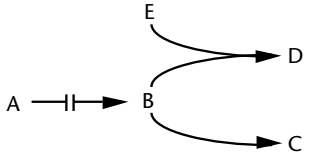
Complex Characters

Polygenic inheritance is usually dealt with in connection with characters that are simple in the sense that they are *measurable*, such as height, body mass, and so on. But of course an anatomical feature such as an eye is an extremely complicated assemblage of smaller structures, each of which may be itself made up of smaller parts. It is therefore hardly surprising that an eye, a stomach or a heart is the product of the action of many, probably thousands, of genes, and at the moment it is impossible to disentangle their individual effects. However, *each* contributing gene is inherited according to normal Mendelian principles

Pleiotropy

Pleiotropy occurs where a gene has multiple effects. Though a gene has a single *primary* effect – the production of a polypeptide – the polypeptide produced often interacts with other polypeptides to produce secondary or 'ripple' effects:

- The reaction an enzyme catalyses may form a part of several metabolic pathways.
- The product of a reaction may be subsequently used in several other metabolic pathways.



A defect in the enzyme converting A to B would also stop the production of D and C.

Many apparently trivial characteristics, such as the ability to roll the tongue or to taste PTC, are probably pleiotropic effects of alleles whose main function is quite different. It is likely that most (if not all) genes have pleiotropic effects. Some pleiotropic effects are complex.

Example

In **sickle-cell anaemia** the primary effect of the sickle haemoglobin gene is a defective β-chain in haemoglobin.

This causes the haemoglobin in homozygotes to crystallise at low oxygen levels in capillaries. As a result, the red cells become sickle-shaped, obstructing the capillaries and causing reduced blood flow.

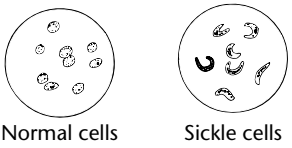


Fig. 8.3: *Sickle cells.*

The effects of impaired circulation are widespread:

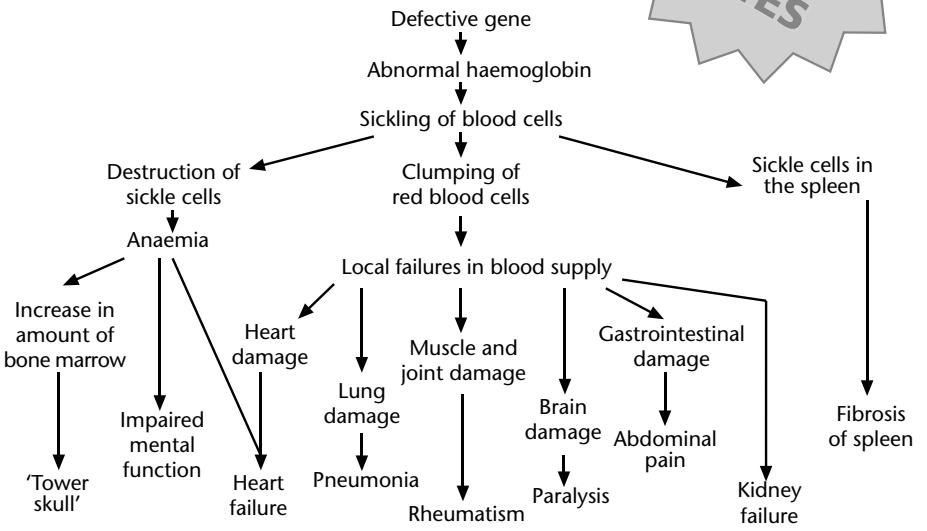


Fig. 8.4: *Pleiotropic effects in sickle-cell anaemia.*

Extension: Pleiotropy and Evolution

One of the problems in evolutionary theory is how new structures originate. Natural selection can only act when structures are already sufficiently well developed to be functional, so how did the fins of fish and the feathers of birds arise in the first place? The Lamarckian explanation ('because there was a need for them') will not do.

One possibility is that structures originate as pleiotropic effects.

Examples

A tiny fold of skin in the body wall of a fish may in the first instance have arisen as a 'by-product' of genes that were concerned with other functions. Even a very slight effect on stability would then be selected for and an increase in size in such outgrowths would have similarly been selected for.

Feathers may similarly have arisen as tiny outgrowths of the skin, pleiotropic effects of genes concerned with functions unrelated to body temperature and flight. Though small outgrowths would have had no significance in flight, they would have significantly improved heat retention and thus could be selected for – flight would have come later.

Activity 8: Polygenic inheritance and pleiotropy

1. **a**. Distinguish between continuous and discontinuous variation, and give an example of each.

b. Which is easier to use in the study of inheritance, continuous or discontinuous variation, and why?

2. Human reflex time is probably controlled by many genes. Explain why.

3. The table shows the results of an experiment involving a cross between two pure-breeding varieties of corn – one short-eared (P_1) and the other long-eared (P_2). The figures below show the frequency distribution of ear length in the parental, F_1 and F_2 generations.

Length of ear (cm)

	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21
P_1	8	22	26	9	1											
P_2							2	10	13	17	27	18	11	6	3	
F_1					1	11	13	16	15	9	3					
F_2			1	10	18	25	42	76	67	61	41	28	14	8	1	

a. What evidence is there in the data to suggest incomplete dominance in these genes?

b. Is the variation in the parental generations likely to be due mainly to genetic or environmental variation? Justify your answer.

c. Which of the generations is likely to show the greatest *genetic* variation?

Page Break

Activity 8: Polygenic inheritance and pleiotropy

1. **a**. See text.

b. Discontinuous inheritance. Because only one gene is involved it is easy to 'quantify' the effects of the genes (***M***). *In comparison*, continuous inheritance involves the interactions of many genes and it is very difficult to separate out individual effects of each gene (***E***).

2. Because reflex times show a range of variation amongst individuals, indicating that several genes are involved. (***M***)

3. **a**. The F_1 are roughly intermediate between the two parents. (***A***)

b. Since the two parents were pure-breeding they were homozygous for the traits. Hence the variation must have been mainly due to environmental causes. (***M***)

c. Since the parental strains were homozygous, each would have produced only one kind of gamete. The F_1 would therefore be genetically uniform but each would be heterozygous for many loci. The F_1 plants would therefore produce many kinds of gamete, and with random fertilisation, many new gene combinations would be produced (***M***). This is supported by the fact that the F_2 plants show greater variation in ear length than the F_1 or either of the two parental strains (***E***).

Glossary/Index

abiotic factor: (5, 167) physical factors in the environment such as temperature, light intensity, humidity.

abscisic acid (**ABA**)**:** (196) plant growth substance that acts as a powerful growth inhibitor and plays an important part in maintaining dormancy in seeds and winter buds, and may also be involved in gravitropism in roots.

abscission: (197) the breaking off of leaves, fruits and flowers as a result of the formation of a layer of weakness at the base.

absorption spectrum: (194) graph of the proportion of light absorbed by a pigment plotted against wavelength.

acheulean: (398) type of tools made by *H. erectus* and *H. heidelbergensis*.

action spectrum: (193) graph in which the effectiveness of light in inducing a process is plotted against wavelength.

active site: (108) part of the surface of a protein in which the specific catalytic properties of the protein are localised.

adaptive radiation: (309) relatively sudden appearance and proliferation of new forms from a single ancestral type to fill a variety of niches.

adenine: (115) one of the four kinds of nitrogenous base found in DNA and RNA.

afferent nerve fibre: (169) nerve fibre that conveys impulses from a receptor to the central nervous system.

agarose: (322) jelly-like material used to separate DNA fragments of different sizes.

***Agrobacterium*:** (345) common soil bacterium used to insert plasmids into flowering plant cells.

allele: (53) one of two or more forms a gene at a given locus can take.

allelopathy: (235) inhibition of the growth of competing species of plants by the production of growth-inhibiting substances by a plant.

allopatric: (287, 298) occupying geographically separate ranges.

allopatric speciation: (298) speciation involving a period of geographical separation.

allopolyploidy: (305) polyploidy resulting from contribution of chromosomes from two or more species.

altricial young: (287) young born in a helpless state, usually blind and with limited powers of movement.

amino acid: (103, 104) one of the subunits from which proteins are built.

amyloplast: (226) starch-storing organelle, probably involved in gravitropism.

aneuploidy: (157) variation in chromosome number involving less than the whole set.

anneal: (321) joining of complementary nucleotide sequences by controlled lowering of temperature.

annual rhythms: (176) rhythms associated with seasonal cycles over a period of 365.25 days.

antibiosis: (236) inhibition of the growth of bacteria by the production of growth-inhibiting substances (antibiotics) by fungi.

antibody: (368) protein produced by the body in response to the presence of a foreign substance or antigen.

anticodon: (131) group of three tRNA bases which is complementary to, and therefore can combine with, an mRNA codon.

antigen: (368) substance which, when introduced into the body, stimulates the production of an antibody which can bind to it.

apical meristem: (220) region of actively-dividing cells at stem or root apex.

assortment: (41) arrangement of chromosomes on the equator of the spindle during meiosis I.

Australopithecus: (385) genus of fossil African prehumans, one species of which was probably ancestral to *Homo*.