GENETICS

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AKHAND PUBLISHING HOUSE
DELHI (INDIA)

Published by

AKHAND PUBLISHING HOUSE

Publisher, Distributor, Exporter having an Online Bookstore

Head Office : L-9A, First Floor, Street No. 42,
Sadatpur Extension, Delhi-110094 (INDIA)
Phone No.: 9968628081, 9555149955 & 9013387535
E-mail : akhandpublishinghouse@gmail.com,
akhandpublishing@yahoo.com
Website : www.akhandbooks.com

GENETICS

© Authors
Ist Edition 2020
ISBN 978-81-945367-7-2

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Printed in India

Published by Jhapsu Yadav for Akhand Publishing House. Cover Designed and Laser Typesetting at VM Graphic and Printed at Aarna Enterprises, Delhi.

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First Author of This Book
Dedicated to
Beloved Father Late Tulsidas Dubey

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Preface

Genetics arose out of the identification of genes, the fundamental units responsible for heredity. Genetics may be defined as the study of genes at all levels, including the ways in which they act in the cell and the ways in which they are transmitted from parents to offspring. Modern genetics focuses on the chemical substance that genes are made of, called deoxyribonucleic acid, or DNA, and the ways in which it affects the chemical reactions that constitute the living processes within the cell. Gene action depends on interaction with the environment.

Mendel's studies have provided scientists with the basis for mathematically predicting the probabilities of genotypes and phenotypes in the offspring of a genetic cross. But not all genetic observations can be explained and predicted based on Mendelian genetics. Other complex and distinct genetic phenomena may also occur. Several complex genetic concepts, described in this section, explain such distinct genetic phenomena as blood types and skin colour.

The human genome is the term used to describe the total genetic information (DNA content) in human cells. It really comprises two genomes: a complex nuclear genome which accounts for 99.9995% of the total genetic information, and a simple mitochondrial genome which accounts for the remaining 0.0005%. The nuclear genome provides the great bulk of essential genetic information, most of which specifies polypeptide synthesis on cytoplasmic ribosomes. Mitochondria possess their own ribosomes and the few polypeptide-encoding genes in the mitochondrial genome produce mRNAs which are translated on the mitochondrial ribosomes. However, the mitochondrial genome specifies only a very small portion of the specific mitochondrial functions; the bulk of the mitochondrial polypeptides are encoded by nuclear genes and are synthesized on cytoplasmic ribosomes, before being imported into the mitochondria.

The diverse genes of any chromosome are designated, so far as they have received names, by the name of one of the modified characters which it helps

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to produce. Thus the gene yellow, near the upper or left end of the X-chromosome of Drosophila, produces in its mutated condition the modified body programmes yellow; while in its unmodified condition it produces the normal grey programmes of the wild fly. In the maps of all the names apply to the modified or mutated condition of the gene.

Regulation of gene expression refers to the control of the amount and timing of appearance of the functional product of a gene. Control of expression is vital to allow a cell to produce the gene products it needs when it needs them; in turn this gives cells the flexibility to adapt to a variable environment, external signals, damage to the cell, etc.

Genome projects are scientific endeavours that ultimately aim to determine the complete genome sequence of an organism (be it an animal, a plant, a fungus, a bacterium, an archaean, a protist or a virus). The genome sequence for any organism requires the DNA sequences for each of the chromosomes in an organism to be determined. For bacteria, which usually have just one chromosome, a genome project will aim to map the sequence of that chromosome. Humans, with 22 pairs of chromosomes and 2 sex chromosomes, will require 24 separate chromosome sequences in order to represent the completed genome. The Human Genome Project was a landmark genome project and some have argued that the era of genomics is one of the more fundamental advances in human history.

The structure of DNA encodes all the information every cell needs to function and thrive. In addition, DNA carries hereditary information in a form that can be copied and passed intact from generation to generation. A gene is a segment of DNA. The biochemical instructions found within most genes, known as the genetic code, specify the chemical structure of a particular protein. Proteins are composed of long chains of amino acids, and the specific sequence of these amino acids dictates the function of each protein. The DNA structure of a gene determines the arrangement of amino acids in a protein, ultimately determining the type and function of the protein manufactured.

This book is useful to researchers, molecular biologists, biotechnologists and teachers engaged in advanced genetics.

—Dr. Ashwani Kumar Dubey

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1

Heredity and Genetic Materials

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HEREDITY

For much of human history people were unaware of the scientific details of how babies were conceived and how heredity worked. Clearly they were conceived, and clearly there was some hereditary connection between parents and children, but the mechanisms were not readily apparent. The Greek philosophers had a variety of ideas: Theophrastus proposed that male flowers caused female flowers to ripen; Hippocrates speculated that "seeds" were produced by various body parts and transmitted to offspring at the time of conception, and Aristotle thought that male and female semen mixed at conception. Aeschylus, in 458 BC, proposed the male as the parent, with the female as a "nurse for the young life sown within her".

Dutch microscopist Anton van Leeuwenhoek discovered "animalcules" in the sperm of humans and other animals. Some scientists speculated they saw a "little man" (homunculus) inside each sperm. These scientists formed a school of thought known as the "spermists". They contended the only contributions of the female to the next generation were the womb in which the homunculus grew, and prenatal influences of the womb.

An opposing school of thought, the ovists, believed that the future human was in the egg, and that sperm merely stimulated the growth of the egg. Ovists thought women carried eggs containing boy and girl children, and that the gender of the offspring was determined well before conception. Pangenesis was an idea that males and females formed "pangenes" in every organ. These pangenes

subsequently moved through their blood to the genitals and then to the children. The concept originated with the ancient Greeks and influenced biology until little over 100 years ago. The terms "blood relative", "full-blooded", and "royal blood" are relicts of pangenesis. Francis Galton, Charles Darwin's cousin, experimentally tested and disproved pangenesis during the 1870s.

Blending theories of inheritance supplanted the spermists and ovists during the 19th century. The mixture of sperm and egg resulted in progeny that were a "blend" of two parents' characteristics. Sex cells are known collectively as gametes (*gamos*, Greek, meaning marriage). According to the blenders, when a black furred animal mates with white furred animal, you would expect all resulting progeny would be gray (a colour intermediate between black and white). This is often not the case.

Blending theories ignore characteristics skipping a generation. Charles Darwin had to deal with the implications of blending in his theory of evolution. He was forced to recognize blending as not important (or at least not the major principle), and suggest that science of the mid-1800s had not yet got the correct answer. That answer came from a contemporary, Gregor Mendel, although Darwin apparently never knew of Mendel's work.

THE MONK AND HIS PEAS

An Austrian monk, Gregor Mendel, developed the fundamental principles that would become the modern science of genetics. Mendel demonstrated that heritable properties are parceled out in discrete units, independently inherited. These eventually were termed genes.

Mendel reasoned an organism for genetic experiments should have:

- A number of different traits that can be studied
- Plant should be self-fertilizing and have a flower structure that limits accidental contact
- Offspring of self-fertilized plants should be fully fertile.

Mendel's experimental organism was a common garden pea (*Pisum sativum*), which has a flower that lends itself to self-pollination. The male parts of the flower are termed the anthers.

They produce pollen, which contains the male gametes (sperm). The female parts of the flower are the stigma, style, and ovary. The egg (female gamete) is produced in the ovary. The process of pollination (the transfer of pollen from anther to stigma) occurs prior to the opening of the pea flower. The pollen grain grows a pollen tube which allows the sperm to travel through the stigma and style, eventually reaching the ovary.

The ripened ovary wall becomes the fruit (in this case the pea pod). Most flowers allow cross-pollination, which can be difficult to deal with in genetic

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studies if the male parent plant is not known. Since pea plants are self-pollinators, the genetics of the parent can be more easily understood. Peas are also self-compatible, allowing self-fertilized embryos to develop as readily as out-fertilized embryos.

Mendel tested all 34 varieties of peas available to him through seed dealers. The garden peas were planted and studied for eight years. Each character studied had two distinct forms, such as tall or short plant height, or smooth or wrinkled seeds. Mendel's experiments used some 28,000 pea plants.

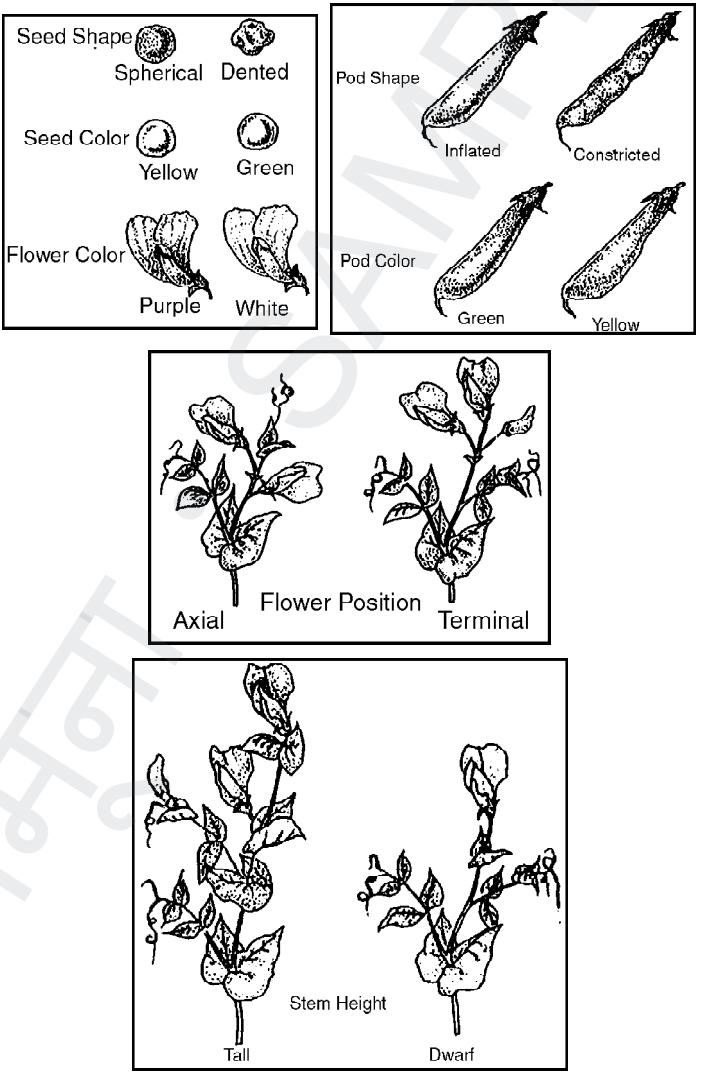


Fig. Some of Mendel's Traits as Expressed in Garden Peas.

Mendel's contribution was unique because of his methodical approach to a definite problem, use of clear-cut variables and application of mathematics