THE WORK OF GREGOR MENDEL - THE FATHER OF GENETICS.
By
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This year marks the 177th anniversary of the birth of the great Austrian scientist-monk, Gregor Mendel, who is venerated as the father of modern genetics.

Mendel was born in 1822 of peasant parents in Moravia (now part of Czechoslovakia). He displayed a great love of learning in school. He studied for a short period at the Institute of Olmutz, but had to leave for financial reasons. He became an Augustinian monk at the monastery at Brno. He studied mathematics and natural history in Vienna to become a schoolmaster teaching science in high school in Brno. In his spare time he studied the results of crossing different varieties of garden peas.

Mendel's interest in plant breeding had been sparked by observations of the results of the hybridisation of ornamental plants to produce new varieties. He noted the regularity of the results and he wondered what would happen if the hybrids were crossed to produce further generations. Garden peas were available in pure-breeding varieties. They reproduce sexually and the reproductive organs are enclosed within the petals so that self-pollination (self-fertilisation) normally takes place. However, the plants can easily be artificially cross-pollinated. Mendel studied 22 different varieties in experiments extending over 8 years.

To illustrate some of the results achieved by Mendel let us consider his studies concerning the position of the pea-flowers on the stem. Mendel used 2 varieties of pure-breeding plants. In one variety the flowers always had an axial distribution, i.e. along the main stem. In the other variety the flowers always had a terminal distribution, i.e. bunched at the top of the stem. Mendel cross-pollinated these two varieties and examined the progeny - called the F1 generation. The parent plants are called the P1 generation.

The male and female sex organs of the same plant are enclosed within the flower. The tip of the male organ is called the anther, which produces the male sex cell - the pollen. The pistil (female organ) produces the egg cell. The cross-pollination was carried out as follows. Mendel removed the anthers of an immature flower of one variety (i.e. terminal or axial flowers) and covered the flower with a small bag to prevent stray pollen from other plants from landing. When the pistil was mature, Mendel transferred pollen from the alternative variety and again covered it. Mendel found that all the plants in the F1 generation were of the axial flower variety.

In an effort to find what had happened to the hereditary factor for terminal flowers, Mendel allowed self-pollination of the F1 plants to produce second generation (F2) plants. In this generation he found that some of the plants were of the terminal-flower variety. The ratio of plants in the F2 generation with axial flowers to plants with terminal flowers was 3 to 1 (actually 3.14 to 1).

Mendel reasoned that there are 2 factors in each plant for flower position, but only 1 of the 2 factors is carried by a pollen grain or an egg. When pollination occurs, the number of factors is restored to 2. The pure-breeding P1 plants with axial flowers contains 2 factors for the axial position. The pure-breeding P1 plants with terminal flowers contains 2 factors for the terminal position. The plants in the F1 generation contained both factors, but all of these plants had axial flowers. Therefore the factor for axial position must be dominant over the factor for terminal

position, which Mendel termed a recessive factor.

When F1 generation pollen grains and eggs are formed, some carry the factor for axial flower position, and some the factor for terminal position. These plants were allowed to self-fertilise and this produced a random mixture of the 2 factors. Some seeds get 2 factors for axial position, some get a factor of each kind, and some get 2 factors for terminal flowers. Mendel calculated by mathematical probability that one quarter of F2 plants would be of the first kind, one half of the second kind, and one quarter of the third kind. The factor for axial flowers is dominant, therefore the first 2 groups have axial flowers and only the third group has terminal flowers, i.e. three fourths of the plants should have axial flowers and one fourth should have terminal flowers. Mendel got an experimental ratio of 3.14 to 1, very close to the mathematical expectations.

Apart from flower position, Mendel used 6 other pairs of alternative traits in his experiments (e.g. round and wrinkled seeds, long and short stems, green and yellow peas, etc.) and found that all behaved in the same manner, i.e. gave a 3 to 1 ratio of dominant to recessive traits in the F2 generation. Each of the ratios differed slightly from 3 to 1, but not enough to raise any serious doubts. Mendel studied large numbers of F2 plants, which was very important. Had he used smaller numbers the results might have deviated more from the mathematical expectation. Such results would be difficult to analyse and interpret. In his experiments, across all 7 pairs of traits, Mendel observed 14,889 dominant traits and 5,010 recessive traits in the F2 generation - a ratio of 2.98 to 1, which is as close to a 3 to 1 ratio as you could hope to achieve under experimental conditions where chance assortment is involved.

The modern word for Mendel's factors is genes. Mendel's interpretation for what happens in such a cross is accepted today as the correct analysis not only for plants, but also for animals. Mendel's system of nomenclature was to represent dominant genetic factors using a capital letter for the dominant gene and the same letter in the lower case for the recessive alternative. The letter is the first letter in the trait which is less common. For flower position, T means axial and t means terminal. That nomenclature is still in use today, e.g. h for the haemophilia gene (bleeder's disease) and H for the alternative dominant gene that is associated with normal blood-clotting.

Of course, it has been known from time immemorial that characteristics are inherited from generation to generation, but until Mendel's brilliant work, nobody understood the mechanism of inheritance. Mendel's identification of the fundamental factors that determine an organism's characteristics and how these factors, which we now call genes, can independently sort and segregate from generation to generation was a revolutionary breakthrough.

Mendel reported his results at a meeting of the Association for Natural Research in Brno in 1865 and published the work that same year in a book under the title Treatises on Plant Hybrids. The work was ahead of its time and was not appreciated by the wider scientific community (nor indeed was it widely read). In 1900 there was a famous re-discovery and appreciation of Mendel's work by Carl Correns in Germany, Hugo de Vries in Holland and Erich von Tschermak-Seysenegg in Austria.

Mendel's work laid the firm foundation for the modern science of genetics. The rise of molecular biology this century identified the chemical nature of the gene as DNA and demonstrated how the gene determines characteristics - by controlling and specifying what

proteins are synthesised. We are now in the middle of a spectacular explosion of genetic-based developments - screening for genetic disease, genetic therapy for disease, genetic engineering of organisms to produce various useful products, cloning of organisms, etc.

In 1936, the British mathematician R.A. Fischer analysed Mendel's results and came to the conclusion that they were simply too good to be true! Fischer claimed that the principles of variation statistics would prevent Mendel from achieving experimental segregation ratios so close to the perfect 3 to 1 ratio. In other words, Mendel, or his assistants, had 'touched up' their data to make it conform to theoretical predictions. Much subsequent work however has failed to support Fisher's doubts. Mendel's experiments stand up. He achieved almost perfect results because he was simply that good. Pure genius.

Mendel was disheartened by the lack of response to his published work. He was elected as Abbot of the monastery in 1868. The heavy work-load of running the monastery took him away from his beloved experiments. Shortly before his death in 1883 he said: 'My scientific studies have afforded me great gratification, and I am convinced it will not be long before the whole world acknowledges the results of my work'. Seventeen years later his prediction came true.

Mendel's experiment showing the ratios for green and yellow peas.
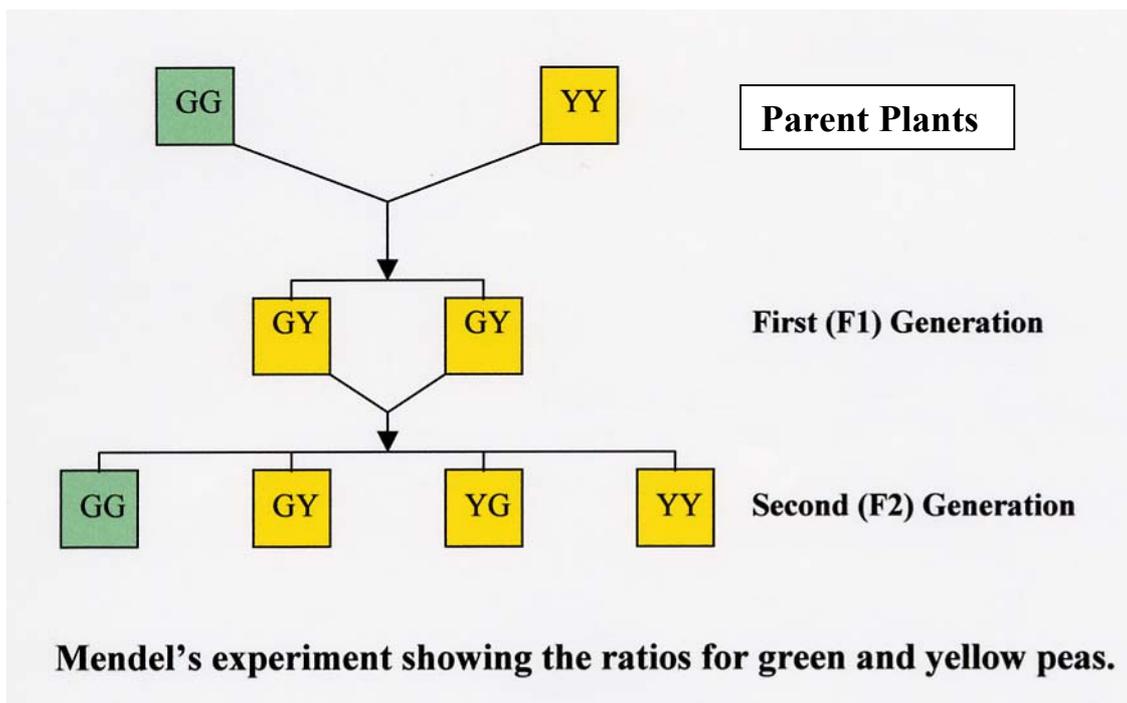
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