MODERN GENETICS 1

Serra

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by

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PREFACE

This is a completely revised and rewritten edition of a book first published in Portuguese about fourteen years ago. In preparing the present edition the author has endeavoured to bring the text up to date with a thorough review of the literature. In so vast a field as genetics, which now embraces biochemistry, microbiology, immunology – as well as cytology and statistics – it is difficult to keep abreast of the enormous output of literature on genetics now appearing. I believe, however, that I have done justice to such work as was accessible and apologize in advance for the inevitable omissions and possible errors. In bibliographical citations, preference has been given to contemporary work rather than to the historical aspect.

The book in its Portuguese version has been successfully used as an advanced genetics text. It is my hope that this new edition may be equally useful to a greater audience of English-speaking readers. It would, of course, have been incomparably easier to write a more specialized or concise book, but it would be difficult to justify yet another short text on genetics. A selection of some of the more useful sections from these three volumes provides an introductory text to the subject; the Portuguese version has often been employed for this purpose. As genetics is becoming one of the major biological sciences, a detailed text where the reader may find bibliographical references to further genetic literature seems eminently justified.

Some areas of genetics, especially those bordering biochemistry and microbiology, are advancing so spectacularly that a review of them can become out of date before publication. These topics cannot, however, be omitted; so in these as well as other areas of rapid advance results up to 1962 or 1963, according to the volume of the book, have been included. The delays associated with publication are a familiar story, aggravated here by the need to revise my own English text, and the further task of bringing the revision up to date. To Mrs. E. Lawton Goodman I express my sincerest thanks and due appreciation for the care and competence shown in performing her difficult task and for the improvements she has brought to the text. My thanks are also due to Drs. Anthony Davies and S. Revell for a part of the revision.

The author alone, of course, is responsible for any errors that may have escaped. No one author is likely to find himself at home in the diverse domains of modern genetics; and the alternative of multi-authorship presents its own problems of unity in presentation, sequence of subject matter, duplication, and so on. I feel sure that readers and critics of the present book will appreciate this: such errors as may be brought to my attention will be corrected in any future edition.

One important domain bordering genetics and evolution, usually called population genetics, has been dealt with only briefly: such basic aspects as the consequences of inbreeding, hybridization and the like, appear throughout some chapters. The inclusion of population genetics would have required a further volume, and there are already several good books on this subject.

I am much indebted to the staff of Academic Press for their assistance with the numerous editorial and production problems. My deep grati-tude is due to Dr. R. M. Albuquerque for her continued and unselfish help with the typescript, bibliography and many of the figures of the book. For further figures, the help of Drs. M. M. P. Seixas, M. M. J. Vicente and P. Picciochi is also acknowledged.

For permission to reproduce figures, I am indebted to many colleagues who, in several cases, were kind enough to send me copies of the figures and new photographs. Due acknowledgement for permission to repro-duce figures is mentioned in the legend of each figure; I hope that authors and publishers will forgive their not being cited individually here. For permission to reproduce some tables of the Appendices to two of the volumes I must mention my indebtedness to the late Sir Ronald A. Fisher and Messrs. Oliver and Boyd, Edinburgh; to the late Mr. W. L. Stevens and the Editor of *Journal of Genetics*, Professor J. B. S. Haldane; also to Iowa College Press.

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Part I: Genetic Transmission and Mendelism

GENETIC TRANSMISSION AND FUNDAMENTAL CONCEPTS OF GENETICS

1.1 Historical Note

THE Science of Genetics as it is now understood began in 1900 when Tschermak, in Austria, de Vries, in the Netherlands, and Correns, in Germany, rediscovered the results which Gregor Mendel, an Augustinian monk, had published in 1866 in the *Proceedings of the Natural History Society of Brünn* (or Brno), nowadays a Czechoslovakian town.[1] There were several reasons for the sad neglect of Mendel's work. At that time biologists centred their interests on the transformation of species rather than on the basis of individual variation. Following the appearance in 1859 of Darwin's "Origin of Species" came the realization that evolution was based upon natural selection, but it was thought that the characteristics of the parents were in some way passed on as a "mixture of bloods" to the offspring and that the individuals of a species naturally showed a variation upon which natural selection exerted its action. Heredity was assumed but not explained.

Among other circumstances that contributed to the neglect of Mendel's work was that the results of his experiments were published in an obscure periodical of limited circulation, although Mendel himself made his work known to some of the most famous biologists of his time—but not, of course, to the younger generation of scientists who were, perhaps, less prejudiced against new ideas. The chief circumstance, however, must have been the fact that Mendel himself was unable to justify the generalization of his conclusions to all of the species that he studied. Eight years of work led to the now classical forty-six-page memoir on pea hybridization, in which Mendel presented an interpretation of his observations in almost exactly the same way as a modern geneticist would; even the terms he employed were very similar to those now used. Following his experiments with *Pisum*, Mendel hybridized

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[1] Mendel (1866, 1870). The bibliography indicated in footnotes is cited by title and publication at the end of Volume 1.

Hieracium species, but the results of these crosses were very different from those obtained between strains of peas. Experiments with *Phaseolus* which Mendel undertook at the same time as his work upon *Pisum*, were also inconclusive. Although Mendel stated that he was beginning further studies of hybridization, no additional results were published. It is known, however, that Mendel was by then having trouble with the administration of the monastery of which he had become the Abbot, and this may have been another factor which prevented the continuation of his investigations.

Failure to obtain clear-cut results with interspecific hybrids had already been recorded by several workers including Kölreuter (1760), Wiegmann (1826), Gärtner, Goss, and Naudin. Naudin (1863) almost reached a correct interpretation when he concluded that the hybrids possessed the hereditary properties of both parents, being mixed forms in whose offspring the characters of the parent species may appear separated. In reality, crosses between species are not appropriate to an exact study of the bases of heredity, because the differences between species generally concern many characters and also, as is now known, interspecific hybrids often show abnormal meiotic divisions. Mendel was fortunate in his selection of experimental material, since the strains of peas he crossed differed only in one or a few clear-cut characters whose inheritance was simple. His method of analysis was, however, strikingly original and laid the foundations of the theory of genetic determinants.

Between 1866 and the rediscovery of Mendel's results, some interesting theoretical contributions to the understanding of heredity appeared, although this domain was then considered of secondary importance to a study of evolution. Weismann clearly showed in 1885–87 the possibility of conceptually separating that which is hereditary from that which is not, thus establishing the distinction between soma- and germ-plasm. Galton (1822–1911), who studied human heredity by statistical methods, summarized his conclusions in the form of two laws which related the characteristics of the individual to those of his or her parents, grandparents, or more distant ancestors and with the mean of the population to which the individual belonged.

Meanwhile, in several fields of biology significant progress was achieved; rapid progress was made in embryology and cytology, led by Wolff and by Schleiden and Schwann, respectively. In 1875 Hertwig, and soon afterwards Strasburger, Fol, and others, discovered the chromosomes in the cell nucleus and studied their division. At the same time the role of the chromosomes in heredity was presumed. Boveri and others then described the reduction division. The parallelism between the behaviour of chromosomes and Mendelian results became clear to

several investigators early in the 20th Century. Correns, Sutton, and Boveri in 1902 formulated the chromosomal theory of heredity, another and important proof of which, in the same year, was McClung's discovery of the sex chromosomes.

From 1900 the young science of heredity, soon to be related with cytology in a common domain called cytogenetics, progressed rapidly and became a central aspect of biology, to which it has given cohesion and unity. Genetical theory is basic to animal and plant improvement and every day becomes more important in medicine and for the understanding of human nature.

It would be difficult to summarize the impressive body of facts discovered by geneticists and cytogeneticists during the last sixty years. Because the history of genetics has still to be written[2] only some of its most striking events will be referred to here. In 1906, Bateson and Punnett discovered linkage, although their interpretation of this phenomenon was not correct. In 1909, Janssens related the cytological phenomena of meiosis with the occurrence of factorial recombination.

Many of the facts of genetics have been obtained from a study of the fruit fly (more properly fruit-fermentation fly) *Drosophila*, whose laboratory rearing was begun by Woodworth and Castle in 1904. After 1910, Morgan and co-workers intensively pursued the genetic study of this small fly which is still highly favoured as a material for genetic experimentation. A special scientific co-operation centre, the Drosophila Information Service (now paralleled by similar services for other species —maize, the mouse, *Neurospora*, *Antirrhinum*, and various microorganisms) summarizes genetic and cytogenetic results obtained by using *Drosophila*. Among other landmarks in genetics are the plotting of the first chromosome map in 1913 by Sturtevant, the studies of Bridges on the chromosomal theory of inheritance, the first experimental production of mutations by X-rays, by Muller, in 1927, and the studies of the Oenotheras by Cleland and Renner.

More recent events that could be cited are the establishment of the correct interpretation of the salivary gland chromosomes of Diptera (by Heitz and his co-worker Bauer in 1933, and soon afterwards by Painter), the demonstration in 1944 by Avery, MacLeod and McCarthy of the nature of bacterial transforming principles discovered by Griffith in 1928, and the finding of transduction in 1952 by Lederberg and Zinder.

The theoretical elaboration of genetic concepts and generalizations begun by Mendel himself, has progressed side by side with the establishment of facts. Reference must be made to Johannsen in whose book

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[2] A historical summary of some genetic discoveries may be found in Babcock (1949).

(1909) the concept of genotype and phenotype was clarified and who coined the word gene[3] (more or less synonymous with the "factor" of Mendel) from which the name of genetics was afterwards derived. Among the theoretical geneticists, Morgan and his disciples, especially Muller, are prominent but the names of Haldane, Goldschmidt, Fisher and Wright must be mentioned too, and even this list probably does little justice to other men of equal merit.

1.2 The Link Between Successive Generations. Heredity and Life Cycles

Many plants show asexual reproduction in which part of the plant is detached and can grow, under suitable conditions, into a complete new individual. Animals of the lower groups frequently multiply asexually. In Protozoa and the plants which may be grouped under the collective name of Protophyta, reproduction by simple division, binary or multiple, is widespread.[4] Individuals derived by asexual reproduction from a single individual, collectively form a *clone*.

More prevalent than asexual multiplication in the majority of plant and animal groups is sexual reproduction, in which typically the bridge from generation to generation is a single cell, the zygote. Sexual reproduction has survival value in that it provides one of the most general and sure mechanisms for *recombination*: this is the process of sorting out and redistributing in different combinations the hereditary determinants of a cell or organism. Recombination, in this generalized sense, is one of the means by which living entities achieve the genetic variation upon which natural selection can work.[5]

The life cycle of a typical metazoan is represented in Fig. 1.1, in which it is supposed that the sexes are separated. Usually the higher animals are diplont; that is, their life cycle occurs practically all in the diploid phase (nuclei with $2n$ chromosomes). Only the secondary spermatocytes, spermatids and spermatozoa in the male, and the corresponding cells in the female (secondary oöcytes and egg cells) are haploid (with n chromosomes).

In Fig. 1.1 a scheme of differentiation into *somaplasm* and *germplasm* is shown, the latter formed by the reproductive cells and their forerunners, the former constituting the remainder of the body of the

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[3] A more recent reprint of this book: Johannsen (1926).

[4] Descriptions of these processes of multiplication are to be found in books of general biology, for instance, Hartmann (1933) or Haupt (1940).

[5] This point is elaborated in Chapter 5.

individual. This differentiation is a special case of germinal localization, whose precocity varies from species to species; for instance, in the small hermaphrodite marine animal *Sagitta*, differentiation is particularly early, a special more stainable body being discernible in the cytoplasm of the fertilized egg. Up to the fourth segmentation division, this body is maintained in a single cell, from which the germinal cells of testes
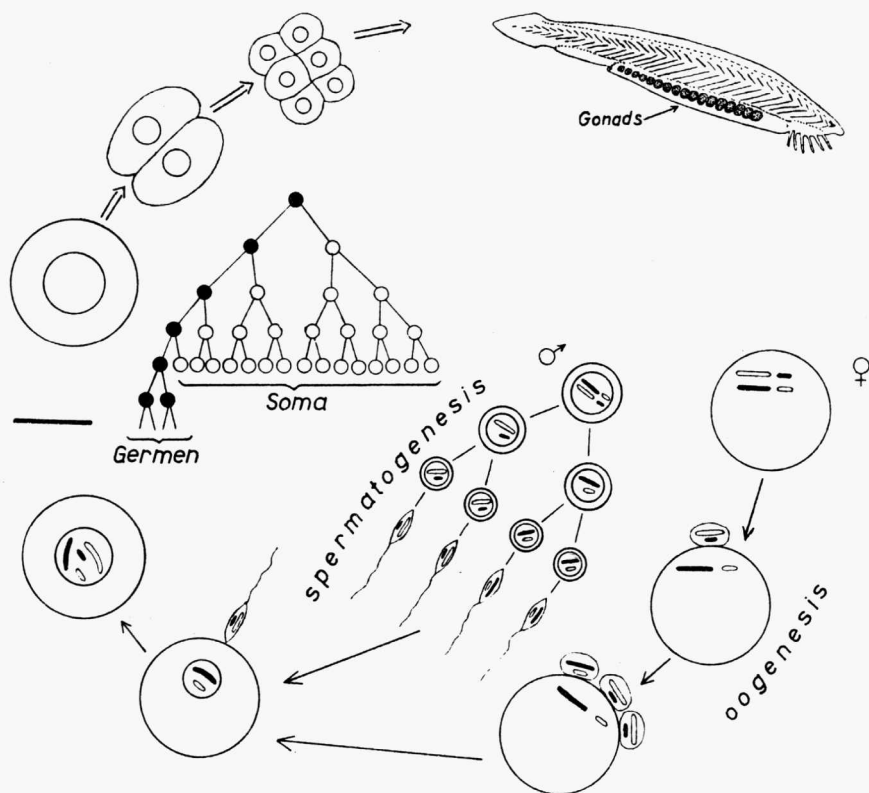

FIG. 1.1. Life cycle of a metazoan with the sexes separated. The diploid chromosome number is assumed to be four. Meiosis takes place in males during the formation of the sex cells, in the passage from primary spermatocytes to secondary spermatocytes and from these to spermatids. In females meiosis occurs in the passage from primary oöcytes to secondary oöcytes and mature egg cells. In spermatogenesis, four spermatids, and from these four spermatozoa, are formed from each primary spermatocyte, while in oögenesis only one egg cell and two or three polar bodies are formed from one primary oöcyte. Fertilization produces a diploid zygote, which, through embryogenesis (only the first segmentation divisions are represented) gives a new individual.

The inset represents a case of precocious differentiation of soma and germ cells. The two cells formed at the first segmentation division of the zygote are supposed to be different, one of them giving rise to the germ line after several mitoses. See text.

and ovaries are formed.[6] In other species differentiation into soma- and germplasm occurs a little later; for instance, it has been assumed to occur in the first segmentation division in *Parascaris equorum* (*Ascaris megalocephala*),[6a] as shown in Fig. 1.1. In some cases where it is possible to regenerate complete organisms from a somatic part of the body, it is concluded that differentiation into soma- and germplasm either does not occur, or that it takes place only during the development of the sexual organs themselves.

The life cycle of a typical monoecious higher plant is represented in Fig. 1.2. The haploid phase is reduced in this case to the macrospore or embryo sac and the microspore or pollen grain, which are the sexual cells equivalent to those of a metazoan and which fuse to form the zygote or fertilized egg.
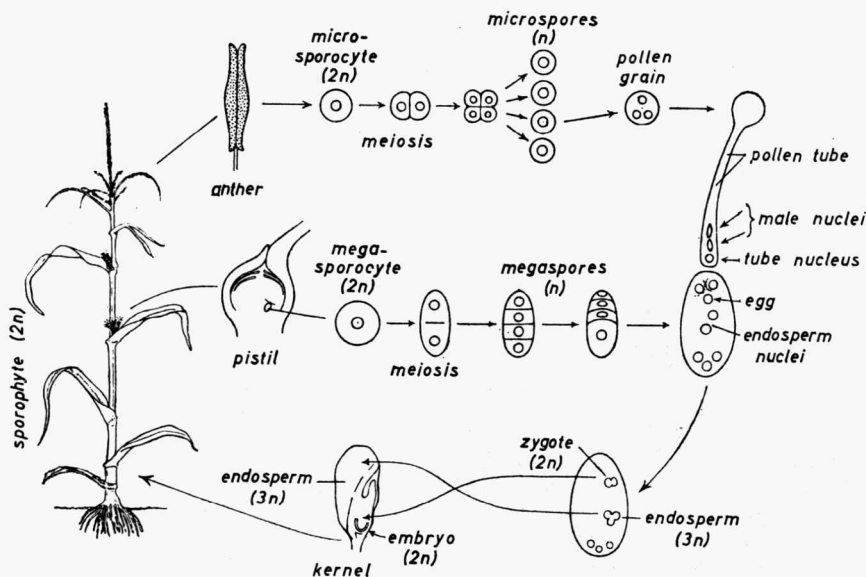

FIG. 1.2. Life cycle of a monoecious angiospermous plant with unisexual flowers. Meiosis takes place in the microsporocytes, each of which produces four microspores, and in the megasporocytes, each of which produces a single megaspore and three abortive cells. The microspore produces the pollen grain and the megaspore the embryo sac. Fertilization is double. The oösphere or egg nucleus fuses with one of the male nuclei of the pollen tube and the zygote forms the diploid (2n) embryo of the seed. The two endosperm nuclei, after fusion with the other male nucleus, multiply to form the nuclei of the endosperm, a triploid (3n) tissue.

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[6] See Buchner (1914).

[6a] Newer observations (Serra and co-workers, to be published) cast some doubt on the current interpretation of early differentiation into soma- and germ-cells in *Parascaris*.

Other types of life cycle occur among animals, including those of many Protozoa and some Metazoa, and in the vegetable kingdom, those of many Algae and Fungi and of the Bryophyta and Pteridophyta.
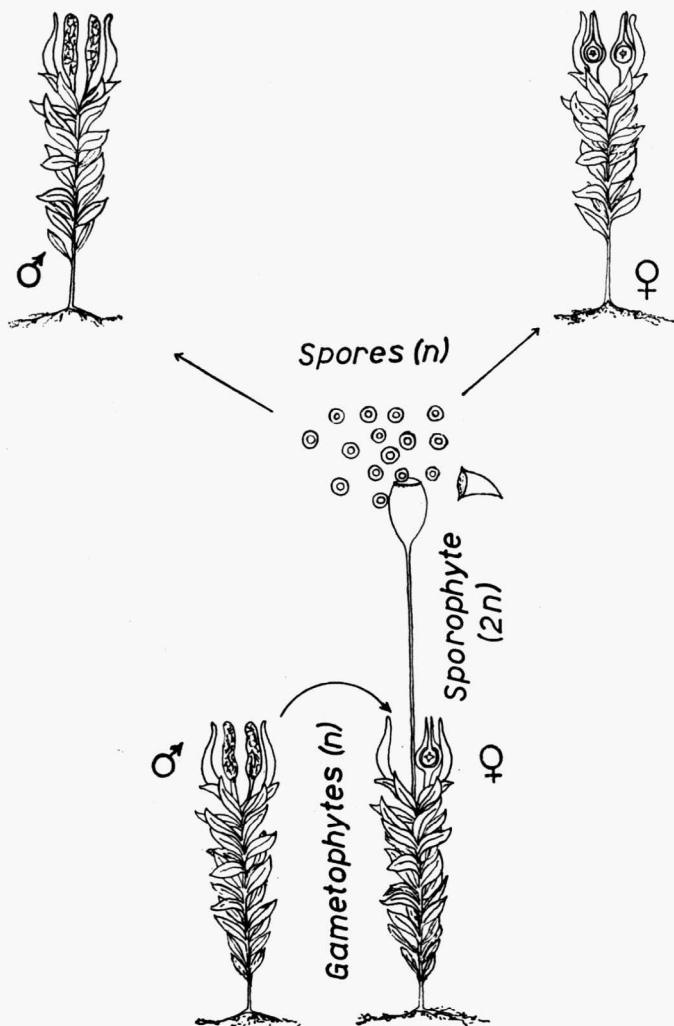

FIG. 1.3. Life cycle of a moss with the two sexes on different plants (dioecious). The plants are haploid gametophytes. Fertilization of the oösphere by a sperm from the antheridia of the male plant produces a diploid zygote, which multiplies to form the diploid sporophyte, constituted by the sporogenous capsule and its stalk, living on the female plant. Haploid spores are produced by meiosis within the capsule. Germination of the spore produces a protonema on which new plants develop. The most developed phase is the haploid gametophyte.

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