INTRODUCTION TO

Genetics and Cytogenetics

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PREFACE

In this book I have endeavored to state and to explain the basic principles of biological inheritance and to show the importance of those principles to man, to the improvement of plants and animals, and to organic evolution. I have attempted to present this material in a simple fashion so that any reader can grasp the fundamentals of heredity in spite of limited biological training. However, I have also included some of the data that support these principles so that the student who wishes can acquire an adequate background for further studies in heredity, and I have added a fairly extensive bibliography so that the more serious student will have a diversified list of some of the items of the research literature should he wish more information on a subject than a book of this size can offer.

Throughout this book I have stressed general principles rather than practical applications and have drawn my illustrations from both the Plant and the Animal Kingdoms. For the reader who is interested in human biology, references to inherited traits are numerous, and Chapters 3 and 19 should be of especial importance. The emphasis on principles and the variety of the illustrations should make this book of value to students of agriculture, psychology, and sociology. It should serve also as a foundation for advanced work in genetics and cytogenetics.

The book is readily divisible into four parts. The first five chapters provide a survey of general biological information which must be understood before progressing into the field of genetics itself. In Chapters 6 through 13 I discuss the fundamental principles of the transmission of genes. In discussing the method by which genes are distributed from generation to generation, I have used the cytological approach, describing chromosomes and their behavior at cell division and reproduction. Chapters 14 through 23 make up the third part of the book. They deal with the nature and physiology of genes and also include some topics of practical and of general interest.

Chapters 24 through 30, the fourth and last part of the book, deal with what are frequently called "chromosomal aberrations." If we accept an ideal concept of chromosomal behavior during cell division and reproduction and if we accept the $2n$ number as the ideal number of chromosomes in the animal soma or in the plant sporophyte and the n number as ideal in the plant gametophyte, any departure from these ideal conditions represents an aberration. The various types of aberrations are described in this section, and their bearing on problems of evolution is discussed. This material is often called "cytogenetics," although any correlation at all between genetic data and cytological observations should properly bear this designation.

Throughout I have tried to avoid being dogmatic on all or most controversial issues. Sometimes I have attempted to present all the important theories concerned in the explanation of certain data without expressing any preference, and on some points where I have favored one theory I have presented other theories for the student to consider.

Because of its scope, I have had to restrict the bibliography somewhat. Many important papers have had to be omitted entirely and where an author had published a series of papers on the same subject, I have listed only a few. Although I did not adhere rigidly to any rule, I frequently listed the first paper of the series and the most recent. I usually, also, included papers that contained extensive bibliographies or summarized information and those that were especially outstanding for the theories or conclusions that they presented. Even though a paper was referred to in more than one chapter, I included it in the bibliography only once.

Several persons have read all or part of the manuscript, and to them I wish to express my deepest appreciation. However, I must emphasize that they are in no way responsible for any of the errors that may appear in the book. Professor George H. Shull of Princeton University has read and criticized the entire text in manuscript, and I am very grateful to him for many suggestions. I also wish to thank Professor P. W. Whiting of the University of Pennsylvania for his kindness in reading and criticizing the manuscript of parts of Chapters 16 and 29. Doctor Alexander Wiener of Brooklyn, New York, read the manuscript of most of Chapter 19 and made many suggestions

that I greatly appreciate. I am grateful also to Doctor Edgar Anderson of the Missouri Botanical Garden for reading the page proof and for an important suggestion.

Many of the diagrams and illustrations are original, but in any book of a general nature it is necessary to borrow from the published works of others. I am indebted to Professor R. A. Fisher, also to Messrs. Oliver and Boyd, Ltd., Edinburgh, for permission to reprint Table III from their book *Statistical Methods for Research Workers*. I also wish to express my appreciation to the University of Chicago Press for permission to use Figure 10, which had previously appeared in the *Botanical Gazette*, to the *American Naturalist* for permission to borrow Table 20, and to *Scientific Agriculture* for permission to reproduce Table 23. I wish also to express my sincerest thanks to all the numerous journals which gave me permission to use their material, to the many geneticists and cytologists who kindly lent me original drawings or cuts, and to those who gave me permission to redraw their published figures or to reproduce their data. Individual acknowledgments have been made in the legends of the figures or tables.

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CONTENTS

Chapter 1

GENETICS, CELLS, AND CHROMOSOMES

Genetics is one of the numerous branches of the biological sciences. It attempts to discover the laws which determine why certain individuals related by descent resemble one another or why they differ from one another. It is the science of heredity and it attempts to discover how and why certain resemblances "run in families" and why many differences are also found among members of the same family. It is one of the biological sciences for it includes both plants and animals in its investigations, and, especially in its more recent aspects, it borders upon physics and chemistry. It is, furthermore, a relatively new science, not established on a scientific basis before 1900.

The science of genetics is intimately related to another biological science, *cytology*. Cytology is a study of those minute living units, the cells, of which plants and animals are constructed. Among the many structures found in cells are certain bodies, the *chromosomes,* which have been shown to be of the greatest importance to students of genetics because in them are located the hereditary units. In other words, the physical basis for the laws of heredity is to be found in the chromosomes; therefore, a knowledge of cytology or at least of chromosomal cytology is absolutely necessary for an understanding of the principles of the science of genetics.

The intimate relationship between the sciences of genetics and cytology was not realized during their early development. However, as more information became available in both fields of knowledge, a striking parallelism became evident which soon suggested that they were in reality not two separate studies but merely two phases of one. Further experiments only served to corroborate this unity until it became evident that the close relationship between genetics and cytology was incontrovertible.

During the earlier years of scientific investigations into the field of heredity, data were obtained by methods that are con-

sidered purely genetical. When the physical basis of genetic phenomena was realized numerous studies were undertaken using the methods of both genetics and cytology and correlating data obtained by genetic procedures with observations determined by cytological techniques. This dual approach to the problems of heredity has given us the term *cytogenetics*, a term which emphasizes the correlation of information obtained by the two diverse techniques. Many of the methods of cytogenetics make use of chromosomal aberrations, for it is by an intense study of exceptional chromosomal behavior that we obtain our best information in regard to the normal conditions. Although cytogenetics is frequently concerned with aberrations, the term is a broad one and includes all situations in which data from cytology and genetics are studied with reference to each other.

A study of the chromosomes and of their behavior in related species and genera has sometimes aided in a better understanding of the evolutionary relationships of taxonomic groups. Many difficult problems of classification and of relationships have been clarified in whole or in part by supplementing taxonomic studies with those of chromosomal cytology. A study of phylogenetic relationships by the methods of both systematic botany or zoology and chromosomal cytology is sometimes called *cytotaxonomy*.

Resting Cells

As part of the biological background for a study of heredity we must realize that all living organisms are composed of minute structures called *cells*. In the higher animals and plants the body is made up of many cells which may differ greatly in both shape and function.

When a cell is not dividing, it is usually referred to as a *resting* or, more properly, a *metabolic cell*, and it is in this condition that most of the cells of both plants and animals are to be found.

The living part of all cells, whether in the resting stage or dividing, is a very complicated mixture of a number of different chemical substances, known as *protoplasm*. Under the microscope, protoplasm, while alive, appears as a colorless, optically homogeneous fluid containing granules, crystals, and droplets; but, when killed, fixed, and stained, it appears to have a finely

granular nature. In the living condition protoplasm is generally considered to be an emulsion type of colloid consisting of a watery background in which are many tiny globules of an immiscible substance, giving it the appearance of milk that has been shaken up. In the watery part may be suspended many extremely small particles or granules, which may be arranged so as to form an interlacing network. In the liquid part also are various dissolved substances such as salts and sugars. Although protoplasm is generally fluid and has a specific gravity only slightly higher than water, it may sometimes be firmer in consistency than water and more like a jelly.

Protoplasm in all typical living cells can be differentiated into two parts, the *cytoplasm* and the frequently more jelly-like *nucleus*. The outer region of the cytoplasm is firm and membranous and forms the *plasma membrane*. This is of great importance physiologically as it permits some substances to enter and leave the cell and prevents others from doing so. In most plant cells a *cell wall* surrounds the plasma membrane, but this structure is not concerned with the passage of materials into and out of the cell. In young plant cells this wall may be very thin, but in most older cells a thicker secondary wall is also present. In some types of specialized cell this secondary wall becomes very thick.

In the embryonic plant cell (Fig. 1), the space inside the cell wall is occupied by protoplasm. When the cell is not dividing, the nucleus, usually centrally located, is a round or ellipsoidal mass of protoplasm separated from the cytoplasm by the *nuclear membrane*, a barrier that may separate nuclear and cytoplasmic material. In the mature, unspecialized type of plant cell known as a parenchyma cell, a large *vacuole* is present in the center of the cell and the cytoplasm is mostly restricted to the periphery. In the cytoplasm may be found living structures, such as the *plastids* and *chondriosomes* or *mitochondria*, and many nonliving substances, including starch grains, protein granules, droplets of fat or oil, and various crystals.

In a typical animal cell there is no large central vacuole, and in the cytoplasm are chondriosomes and secreted granules, but no plastids. Lying in the cytoplasm to one side of the nucleus is the centrosome, a structure intimately connected with cell

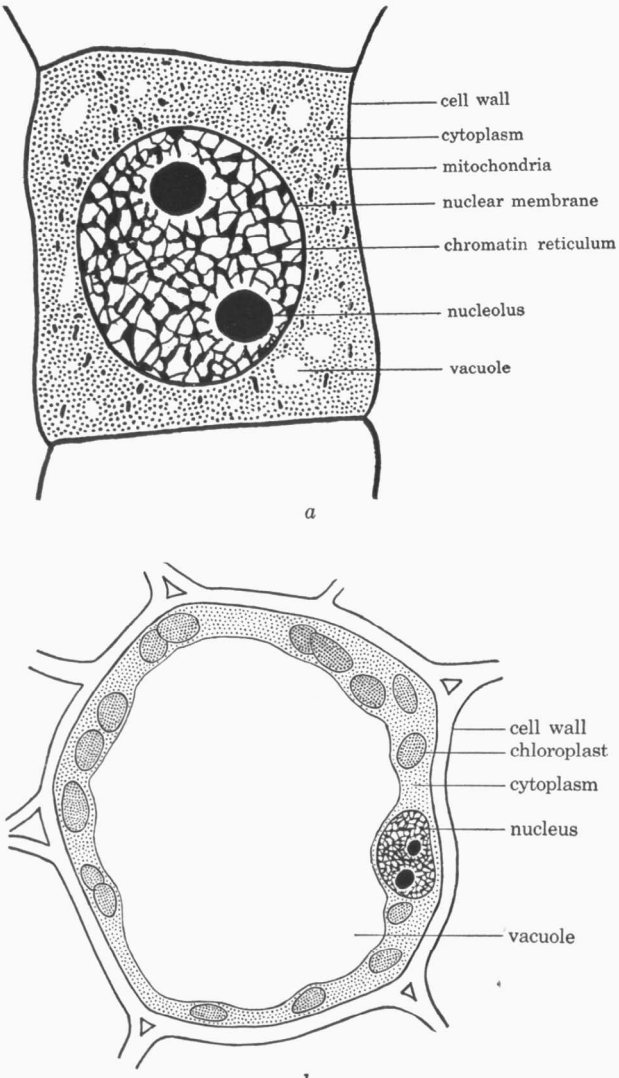

FIG. 1. Plant cells. (*a*) An embryonic cell from the root tip of an onion. ×2800. (*b*) A parenchyma cell from the mesophyll of a leaf of English ivy. ×1400. Camera lucida drawings.

division. This structure, characteristic of animal cells, is also found in some of the lower plants. The centrosome consists of a minute granule, the *centriole,* surrounded by a small mass of protoplasm, the centrosphere; the protoplasm of the centrosphere is often denser than the surrounding cytoplasm. During some stages of division, star-like radiations extend outward from the centrosome into the cytoplasm, forming the *aster.*

Another structure characteristic of animal cells is the *Golgi apparatus.* It is found in the cytoplasm and frequently appears to be a system of connecting canals, but it may sometimes have a more dispersed aspect. Its function is unknown and, although it is characteristic of animal cells, it may, according to some botanists, also be present in some plant cells. No cellulose wall is present in animal cells.

Resting Nucleus

For a geneticist, the most important part of a cell is the nucleus, because in the nucleus are found the genes which determine the characteristics of the organism.

The nucleus is separated from the cytoplasm by a definite membrane, the *nuclear membrane.* The reality of this structure has been shown by microdissection studies. There is good evidence that this membrane is differentially permeable, as is the plasma membrane. If so, the substances to which it is impermeable may be very different from those which will not pass through the plasma membrane.

The structures inside the nuclear membrane are not easily observed in the living condition. Living nuclei generally appear clear and homogeneous, but sometimes seem to consist of many fine granules. Discerning definite structures in the nucleus is difficult because, while alive, most of the structures of a cell are colorless and have almost the same indices of refraction. Also the threads which we know to be present in the resting nucleus are extremely fine and attenuated and are, therefore, more difficult to see than during division stages, when they are many times thicker.

The structures of the nucleus are best observed if the cell is killed, fixed, and stained. By "fixing" is meant treating the cell with certain chemicals that not only kill it but also preserve the cell structures in a condition resembling a living cell. A cell

treated in this manner is readily stained by certain dyes, some of which stain one part of the cell and not others. The parts so stained stand out in marked contrast to the rest of the cell, and their structure is much more easily observed than it is in the living condition.

In the resting nucleus is always found the *karyolymph* or *nuclear sap,* a clear fluid consisting mainly of proteins. In fixed and stained nuclei, the nuclear membrane is generally stained but the nuclear sap appears as an unstained or very lightly stained background inside the membrane. Superimposed on this background are the *chromatin reticulum* and one or more *nucleoli,* both generally stained very deeply.

In fixed and stained slides, the chromatin reticulum has the appearance of a network and is composed of numerous very long and extremely thin threads, in loose coils. These threads are the *chromonemata.* When the cell divides another substance, the *matrix,* condenses on these threads and the chromonemata and matrix together form the *chromosomes,* the most important nuclear structures for the geneticist as they contain the genes.

In the resting nucleus, the chromosomes are individually not distinguishable, but they become identifiable as the cell divides. During cell division it is clear that they exist in definite numbers which are the same not only for all the cells of a given plant or animal, excluding certain reproductive cells, but also, with certain exceptions, for all the individuals of the same species. For example, all the *somatic* (that is, body or nonreproductive) cells of the fruit fly, *Drosophila melanogaster,* normally have 8 chromosomes, whereas those of man have 48. Similarly, cells of all normal maize plants have 20 somatic chromosomes, cells of the garden pea have 14, and those of the onion have 16.

Division of Plant Cells

All cells come from preexisting cells by division. The term *cell division* includes the division of both the nucleus and the cytoplasm, either of which may divide even if the division of the other does not occur. The division of the nucleus is called *mitosis* or *karyokinesis* and the division of the cytoplasm *cytokinesis,* but the use of *mitosis* for the entire process is not unknown. It is customary to divide mitosis into four or five steps which mark definite turning points in the process. Accordingly,

these five steps are frequently recognized: prophase, prometaphase, metaphase, anaphase, and telophase.

Prophase. During the resting or metabolic stage, the chromosomes are so long and thin and so intertwined that they cannot be counted, and there is evidence that each is a single thread until the cell is about to begin to divide (Fig. 2). With the beginning of mitosis, however, a series of profound changes in the nature of the nucleus is begun. There are a shortening and thickening of the chromosomes and a probable loss of water and increase in staining capacity of the individual threads, and if the chromosomes in the resting nucleus are connected by branches or anastomoses, as is frequently believed, these anastomoses are withdrawn at this time. As the result of these changes the individual chromosomes are more readily seen than in the resting nucleus and are no longer joined together in a reticulum. One marked feature of the chromosomes in early prophase is that they are *double* rather than single threads. They appear as two long threads lying parallel and close to one another, each of which contains a specialized region known as the *centromere, kinetochore,* or *spindle fiber attachment point.* In early prophase, the chromosomes are still long and slender and still wind about in a number of loose coils.

As prophase progresses, the chromonemata uncoil and become thicker. The matrix begins to condense on the threads, and the chromosomes at this stage frequently have a fuzzy outline as the result of the irregular accumulation of this deeply staining matrical material along the length of the chromosome.

The two threads that constitute the prophase chromosome are the *chromatids,* each consisting of a chromonema and matrix. The two chromatids are generally visibly uniform throughout except for the centromeres, and the parts on either side of the centromeres are called the *arms.* The region of the arm nearest the centromere is the *proximal region;* the part farthest away is the *distal region.* As prophase progresses, the matrix continues to collect around the chromatids until each chromatid is now a long, rod-like body lying next to its sister chromatid and apparently identical with it in every way. The two centromeres lie side by side and in close contact. As these changes occur in the chromosomes, the nucleolus or nucleoli get smaller and smaller and at about the end of prophase have usually com-

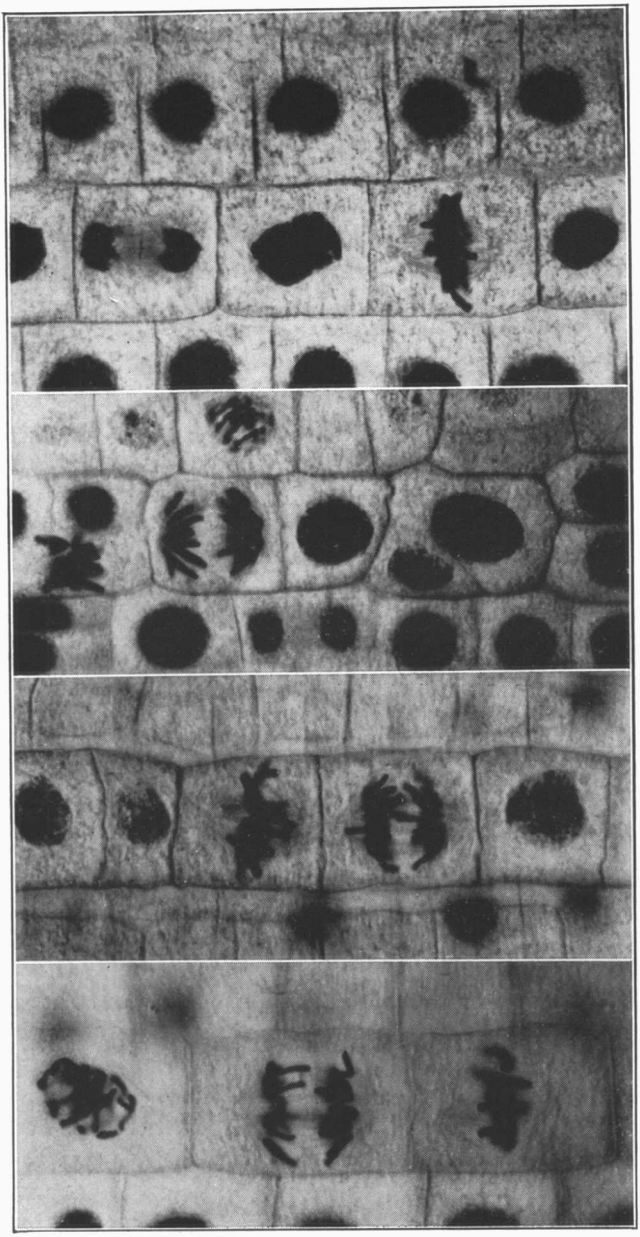

FIG. 2. Photomicrographs of mitosis in onion root tips. *Left*, a typical prophase above, an anaphase in center. *Left center*, typical metaphase in center, anaphase below. *Right center*, late anaphase above. *Right*, early telophase above, metaphase below. (Courtesy Carolina Biological Supply Co.)

pletely disappeared. Towards the end of prophase, the chromosomes have become much shorter and thicker and stain much more deeply than in the earlier stages. They also tend to move towards the outer part of the nucleus. At this time the nuclear membrane dissolves, and with the disappearance of this boundary between the nuclear sap and the cytoplasm, prophase comes to an end.

Prometaphase. When the nuclear membrane disappears, the nuclear sap and cytoplasm are brought into direct contact, and the cytoplasm appears to act upon the nuclear sap so as to cause it to form into a long, spindle-shaped structure known as the *spindle.* In living cells, this structure is not easy to see, but in many fixed and stained cells it appears as a number of fine lines converging to two points. Earlier cytologists believed these lines to be fibers and regarded the spindle as composed of a number of such fibers, which were fairly widely separated in the center of the spindle but converged at the ends. This may be the correct interpretation, but the microdissection studies of Chambers have tended to show that these so-called fibers are not solid.

Whatever is the correct nature of the spindle, it is a firmer, more rigid structure than the cytoplasm in which it is embedded. If the living cells are detached from one another and mounted on a slide, the spindle is crushed only with difficulty, and the cells generally lie so that the spindle is parallel rather than perpendicular to the surface of the slide. The spindle is of great importance in cell division and, if it fails to function properly, mitosis will be abnormal.

The spindle tapers at each end and may or may not come to a sharp point. The ends are called the poles, and the region equidistant between them, the equator. When the spindle has formed, the chromosomes released by the breakdown of the nuclear membrane move towards the equator.

Metaphase. At metaphase, the chromosomes are seen to lie on the equator of the spindle. They frequently arrange themselves so that they lie on the outer part of the spindle with only the centromeres on the equator but sometimes, especially when they are small and numerous, the chromosomes are found in the center as well as in the outer region of the spindle (Fig. 3). The centromeres always lie on the equator, forming an *equatorial*

plate, and the arms often extend away from the equator and may frequently project into the cytoplasm.

The metaphase chromosomes are thick, deeply staining structures. They frequently appear as rod-shaped, V-shaped, or J-shaped bodies, and their particular appearance depends upon the location of the centromere. If it is at the end (*terminal attachment*) the chromosome will appear rod-shaped; if it is at or very near the center (*median or submedian attachment*) it is V-shaped; and if it is near but not at the end (*subterminal attachment*) it has the shape of the letter J. The centromere appears in the metaphase chromosome as a constriction. In addition to the centromere, secondary constrictions may be present near the end and may be very long and deep, so that the end of the chromosome appears as a little knob, called a *satellite* or *trabant.* The function of these secondary constrictions is not well known, but on some chromosomes they are regions at which the chromosome is attached to the nucleolus during the resting stage and from which the nucleolus begins to form at telophase. Each metaphase chromosome still consists of two chromatids but they are very close to one another

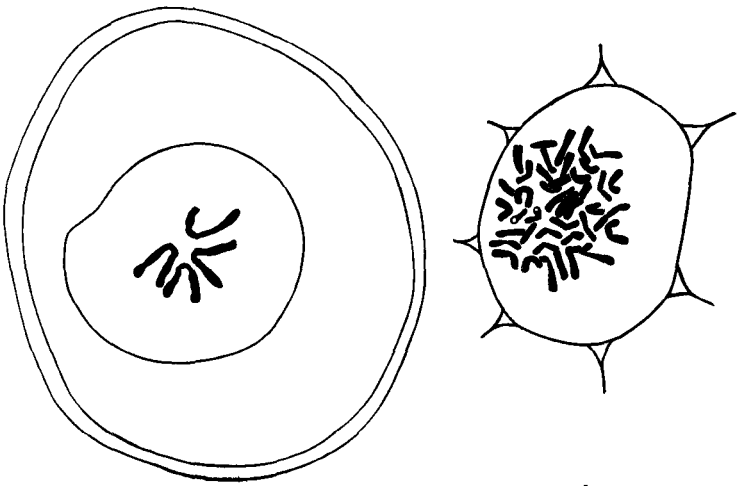

FIG. 3. Polar views of metaphase. (*a*) In the egg of the animal, *Ascaris megalocephala;* ×775. (*b*) In cells of the root tip of *Iris fulva;* ×1500. Camera lucida drawings.