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GENETICS

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SCHAUM'S OUTLINE OF

THEORY AND PROBLEMS

OF

GENETICS

Second Edition

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GENETICS

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Preface

Genetics, the science of heredity, is a fundamental discipline in the biological sciences. All living things are the products of both "nature and nurture". The hereditary units (genes) provide the organism with its "nature" (biological potentialities/limitations); the environment provides the "nurture" which interacts with the genes to give the organism its distinctive anatomical, biochemical, physiological and behavioral characteristics.

Johann (Gregor) Mendel laid the foundation of modern genetics with the publication of his pioneering work on peas in 1866, but his work was not appreciated in his lifetime. The science of genetics began in 1900 with the rediscovery of his original paper. Since then, genetics has rapidly proliferated into numerous distinctive subdisciplines. The kind of studies that Mendel performed are now included in the discipline called Transmission Genetics. Other major areas of investigation are now categorized as Cytogenetics, Cytoplasmic (Plastid) Genetics, Quantitative Genetics, Population (Evolutionary) Genetics, Molecular Genetics, Developmental Genetics and Microbial Genetics. Further specializations are indicated by fields labeled *Drosophila* Genetics, Human Genetics, Fungal Genetics, Viral Genetics, Animal and Plant Breeding, Immunogenetics, etc: This revision touches upon problems in all of these fields without necessarily devoting a complete chapter to each of them.

It has been said that a body of knowledge can be called a science to the degree that its principles can be expressed in mathematical terms. If this is true, then genetics reigns as queen among the biological sciences. The mathematics of elementary genetics is not difficult, but solving genetic problems does require a logical mind and some expertise in working with probabilities. The only mathematical background needed for understanding the concepts in this volume is arithmetic and the elements of algebra. At least some exposure to college biology is desired, but even basic biological principles are reviewed to provide a common base of background information.

The first edition of *Genetics* was developed in 1969 as an aid to students in genetics classes dominated by a mathematical, problem-solving mode of teaching. At that time, this was probably the only such aid available. In the intervening years, other study aids became available, microelectronic calculators became relatively inexpensive, the understanding of basic genetic processes deepened profoundly and a new era of genetic engineering emerged. This second edition reflects the rapid growth of genetic knowledge in the last decade. Extensive reorganization and amplification of information has been made in the areas of DNA replication, protein structure and synthesis, regulatory mechanisms of gene activity, mutagenesis, bacterial and viral genetics, human cytogenetics, quantitative genetics and breeding principles. Three new chapters have been added. Chapter 8 incorporates principles from all of the previous chapters and requires the student to devise solutions to problems without any textual clues as to which principles are being tested. Chapter 13 is an introduction to the quantitation of evolutionary forces. The final chapter presents some of the major technological innovations that have advanced our understanding of genetics at the molecular level and the birth of the age of genetic engineering. Breaking with the tradition established in the first edition, many of the new problems in this revision do not have a mathematical solution, but rather require short essay-type answers. While this book is still primarily intended as a supplemental study aid to any standard genetics textbook, the statements of theory and principle are sufficiently complete for it to be used as a text by itself for a one or two semester course, depending on the desired depth of study.

Each chapter begins with a clear statement of pertinent definitions, principles and background information, fully illustrated by examples. This is followed by sets of solved and supplementary problems. Answers to the supplementary problems are given at the end of each chapter. The solved problems illustrate and amplify the theory, bring into sharp focus those fine points without which

the student continually feels himself or herself on unsafe ground, and provide the repetition of basic principles so vital to effective learning. The supplementary problems serve as a complete review of the material of each chapter.

I am indebted to the Literary Executor of the late Sir Ronald A. Fisher, to Dr. Frank Yates, and to Oliver & Boyd Ltd., Edinburgh, for permission to publish a portion of Table IV from the 6th edition of *Statistical Tables for Biological, Agricultural, and Medical Research.* I am also indebted to many people around the world (the first edition of this book was translated into five languages) who have offered helpful comments over the years since the first printing. I would appreciate receiving further suggestions about this new edition.

WILLIAM D. STANSFIELD

CONTENTS

Chapter 1

The Physical Basis of Heredity

GENETICS

Genetics is that branch of biology concerned with heredity and variation. The hereditary units which are transmitted from one generation to the next (inherited) are called *genes*. The genes reside in a long molecule called deoxyribonucleic acid (DNA). The DNA, in conjunction with a protein matrix, forms nucleoprotein and becomes organized into structures with distinctive staining properties called *chromosomes* found in the nucleus of the cell. The behavior of genes is thus paralleled in many ways by the behavior of the chromosomes of which they are a part. A gene contains coded information for the production of proteins. DNA is normally a stable molecule with the capacity for self-replication. On rare occasions a change may occur spontaneously in some part of DNA. This change, called a *mutation*, alters the coded instructions and may result in a defective protein or in the cessation of protein synthesis. The net result of a mutation is often seen as a change in the physical appearance of the individual or a change in some other measurable attribute of the organism called a *character* or *trait*. Through the process of mutation a gene may be changed into two or more alternative forms called *allelomorphs* or *alleles*.

Example 1.1. Healthy people have a gene which specifies the normal protein structure of the red blood cell pigment called hemoglobin. Some anemic individuals have an altered form of this gene, i.e. an allele, which makes a defective hemoglobin protein unable to carry the normal amount of oxygen to the body cells.

Each gene occupies a specific position on a chromosome, called the gene *locus* (*loci*, plural). All allelic forms of a gene therefore are found at corresponding positions on genetically similar (*homologous*) chromosomes. The word "locus" is sometimes used interchangeably for "gene". When the science of genetics was in its infancy the gene was thought to behave as a unit particle. These particles were believed to be arranged on the chromosome like beads on a string. This is still a useful concept for beginning students to adopt, but will require considerable modification when we study the chemical basis of heredity in Chapter 14. All the genes on a chromosome are said to be *linked* to one another and belong to the same *linkage group*. Wherever the chromosome goes it carries all of the genes in its linkage group with it. As we shall see later in this chapter, linked genes are not transmitted independently of one another, but genes in different linkage groups (on different chromosomes) are transmitted independently of one another.

CELLS

The smallest unit of life is the *cell*. All living things are composed of these basic units, from the simple unicellular structures of bacteria and protozoa to the complex structures of trees and humans. Even within an individual all of the cells do not look alike. A muscle cell is obviously different from a nerve cell which in turn is different from a blood cell, etc. Thus there is no such thing as a typical cell type. Fig. 1-1 below is a composite diagram of an animal cell showing subcellular structures called *organelles* which many types of cells share in common. Most organelles are too small to be seen with the light microscope, but their structure can be studied with the electron microscope. These organelles perform distinctive functions which in total produce the characteristics of life associated with the cell (Table 1.1).

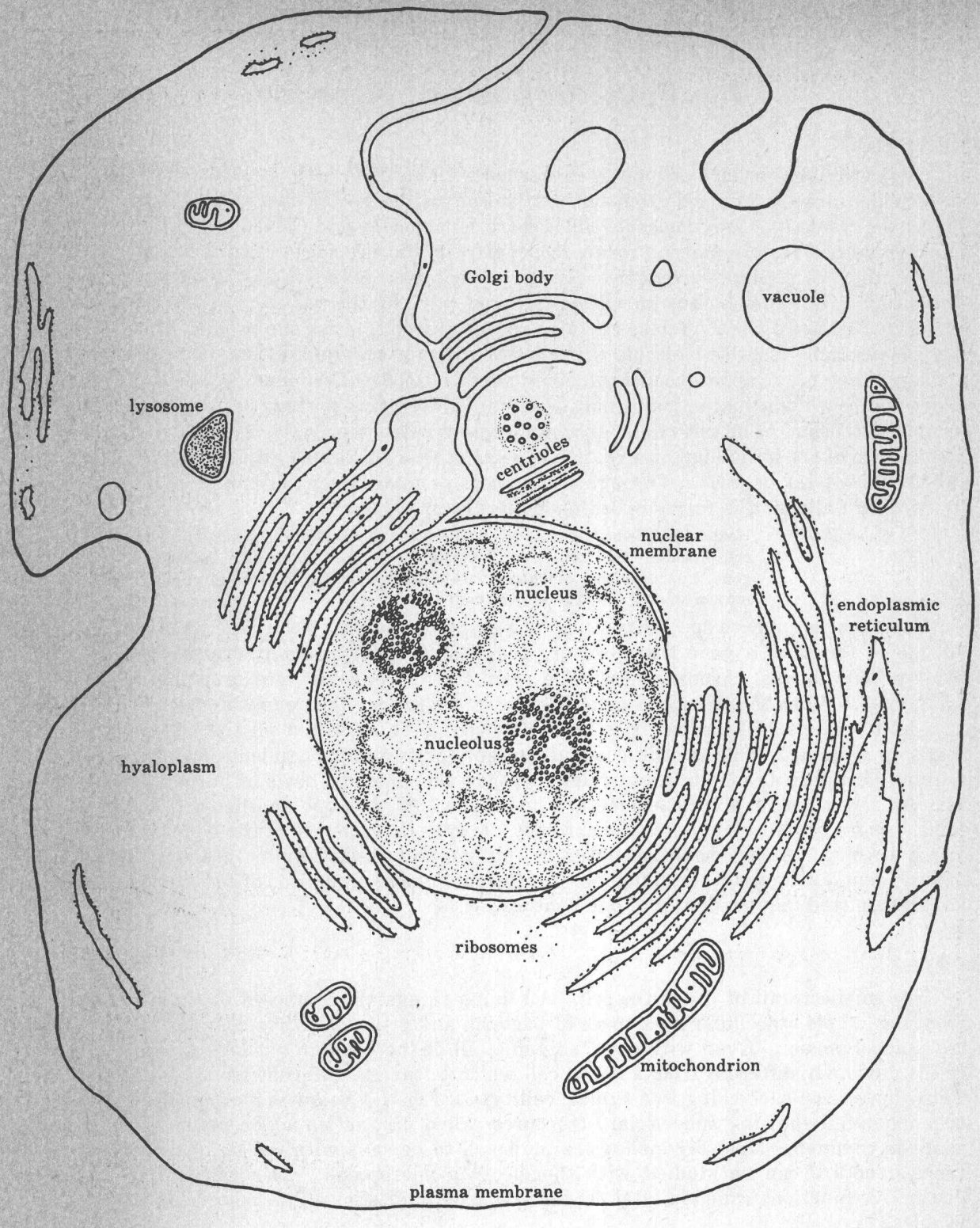

Fig. 1-1. Diagram of an animal cell.

Table 1.1. Functions of Cellular Organelles

Cell Organelle	*Functions*
Cell or plasma membrane	Differentially permeable membrane through which extracellular substances may be selectively sampled and cell products may be liberated.
Cell wall (plants only)	Thick cellulose wall surrounding the cell membrane giving strength and rigidity to the cell.
Nucleus:	Regulates growth and reproduction of the cell.
Chromosomes	Bearers of hereditary instructions; regulation of cellular processes (seen clearly only during nuclear division).
Nucleolus	Synthesizes ribosomal RNA; disappears during cellular replication.
Nucleoplasm (nuclear sap)	Contains materials for building DNA and messenger molecules which act as intermediates between nucleus and cytoplasm.
Nuclear membrane	Provides selective continuity between nuclear and cytoplasmic materials.
Cytoplasm:	Contains machinery for carrying out the instructions sent from the nucleus.
Endoplasmic reticulum	Greatly expanded surface area for biochemical reactions which normally occur at or across membrane surfaces.
Ribosomes	Sites of protein synthesis (shown as black dots lining the endoplasmic reticulum in Fig. 1-1).
Centrioles	Form poles for the divisional processes; capable of replication; usually not seen in plant cells.
Mitochondria	Energy production (Kreb's cycle, electron transport chain, beta oxidation of fatty acids, etc.).
Plastids (plants only)	Structures for storage of starch, pigments, and other cellular products. Photosynthesis occurs in chloroplasts.
Golgi body or apparatus	Production of cellular secretions; sometimes called dictyosomes in plants.
Lysosome (animals only)	Production of intracellular digestive enzymes which aid in disposal of bacteria and other foreign bodies; may cause cell destruction if ruptured.
Vacuoles	Storage depots for excess water, waste products, soluble pigments, etc.
Hyaloplasm	Contains enzymes for glycolysis and structural materials such as sugars, amino acids, water, vitamins, nucleotides, etc. (nutrient soup or cell sap).

CHROMOSOMES

1. Chromosome Number.

In higher organisms, each *somatic* cell (any body cell exclusive of sex cells) contains one set of chromosomes inherited from the *maternal* (female) parent and a comparable set of chromosomes (*homologous* chromosomes or *homologues*) from the *paternal* (male) parent. The number of chromosomes in this dual set is called the *diploid* ($2n$) number. The suffix "-ploid" refers to chromosome "sets". The prefix indicates the degree of ploidy. Sex cells or *gametes*, which contain half the number of chromosome sets found in somatic cells are referred to as *haploid* cells (n). A *genome* is a set of chromosomes corresponding to the haploid set of a species. The number of chromosomes in each somatic cell is the same for all members of a given species. For example, human somatic cells contain 46 chromosomes, tobacco has 48, cattle 60, the garden pea 14, the fruit fly 8, etc. The diploid number of a species bears no direct relationship to the species position in the phylogenetic scheme of classification.

2. Chromosome Morphology.

The structure of chromosomes becomes most easily visible during certain phases of nuclear division when they are highly coiled. Each chromosome in the genome can usually be distinguished from all others by several criteria, including the relative lengths of the chromosomes, the position of a structure called the *centromere* which divides the chromosome into two arms of varying length, the presence and position of enlarged areas called knobs or chromomeres, the presence of tiny terminal extensions of chromatin material called satellites, etc. A chromosome with a median centromere (*metacentric*) will have arms of approximately equal size. A *submetacentric* or *acrocentric* chromosome has arms of distinctly unequal size. If a chromosome has its centromere at or very near one end of the chromosome it is called *telocentric*. Each chromosome of the genome (with the exception of sex chromosomes) is numbered consecutively according to length, beginning with the longest chromosome first.

3. Autosomes vs. Sex Chromosomes.

In the males of some species, including humans, sex is associated with a morphologically dissimilar (*heteromorphic*) pair of chromosomes called *sex chromosomes*. Such a chromosome pair is usually labeled X and Y. Genetic factors on the Y chromosome determine maleness. Females have two morphologically identical X chromosomes. The members of any other homologous pairs of chromosomes (homologues) are morphologically indistinguishable, but usually are visibly different from other pairs (non-homologous chromosomes). All chromosomes exclusive of the sex chromosomes are called *autosomes*. Fig. 1-2 shows the chromosomal complement of the fruit fly *Drosophila melanogaster* ($2n = 8$) with three pairs of autosomes (2, 3, 4) and one pair of sex chromosomes.

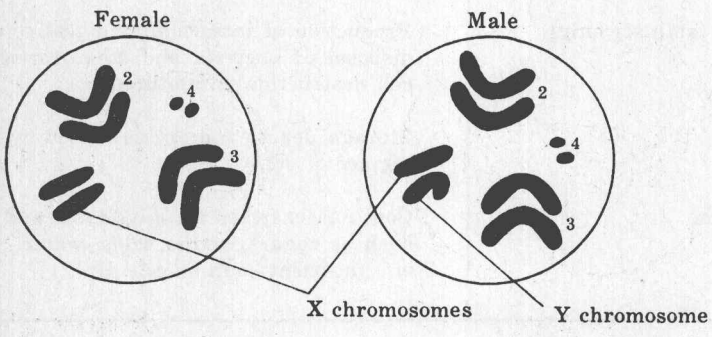

Fig. 1-2. Diagram of diploid cells in *Drosophila melanogaster*.

CELL DIVISION

1. Mitosis.

All somatic cells in a multicellular organism are descendants of one original cell, the *zygote,* through a divisional process called *mitosis* (Fig. 1-3). The function of mitosis is first to construct an exact copy of each chromosome and then to distribute, through division of the original (mother) cell, an identical set of chromosomes to each of the two daughter cells. *Interphase* is the period between division cycles. When the cell is ready to begin mitosis each DNA molecule (which is an integral part of the chromosome) *replicates* or makes an exact copy of itself. This copying process produces a chromosome with two identical functional strands called *chromatids,* both attached to a common centromere. At this stage the chromosomes are in a very long attenuated condition and appear only as chromatin granules under the light microscope. A mitotic division has four major stages: prophase,

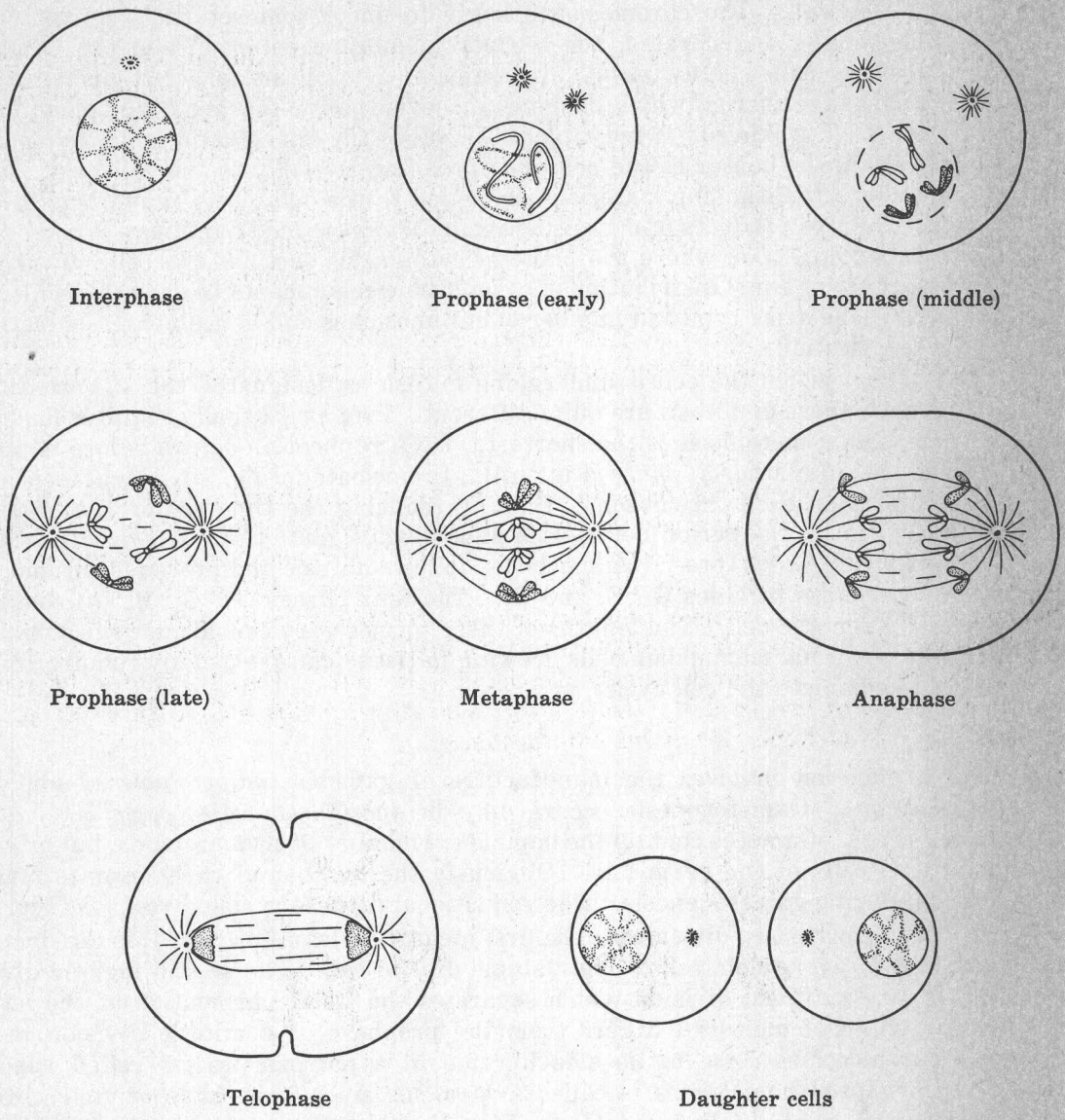

Fig. 1-3. Mitosis in animal cells.

metaphase, anaphase and telophase. In *prophase*, the chromosomes become visible in the light microscope presumably due to coiling, shortening and thickening, and adding protein matrix to their mass as the process continues. By late prophase, the two identical or "sister" chromatids may be seen. The centrioles migrate to opposite ends of the cell and there establish mitotic centers (poles) from which spindle fibers organize and extend to the centromeres. The nuclear membrane begins to degenerate and by *metaphase* is completely gone. Now the centromeres move to the center of the cell, a position designated the equatorial plane or metaphase plate, and the spindle fibers, collectively known as the spindle apparatus, become fully formed. *Anaphase* begins when the centromere splits in two, allowing sister chromatids to separate and move towards opposite poles led by their centromeres. At this stage we may arbitrarily designate the separated sisters as new chromosomes. The arms of each chromosome drag behind their centromeres, giving them characteristic shapes depending upon the location of the centromere. Metacentric chromosomes appear V-shaped, submetacentric chromosomes appear J-shaped, and telocentric chromosomes appear rod shaped. In *telophase*, an identical set of chromosomes is assembled at each pole of the cell. The chromosomes begin to uncoil and return to an interphase condition. The spindle degenerates, the nuclear membrane reforms, and the cytoplasm divides in a process called *cytokinesis*. In animals, cytokinesis is accomplished by the formation of a cleavage furrow which deepens and eventually "pinches" the cell in two as shown in Fig. 1-3. Cytokinesis in most plants involves the construction of a *cell plate* of pectin originating in the center of the cell and spreading laterally to the cell wall. Later, cellulose and other strengthening materials are added to the cell plate, converting it into a new cell wall. The two products of mitosis are called *daughter cells* and may or may not be of equal size depending upon where the plane of cytokinesis sections the cell. Thus while there is no assurance of equal distribution of cytoplasmic components to daughter cells, they do contain exactly the same type and number of chromosomes and hence possess exactly the same genetic constitution.

The time during which the cell is undergoing mitosis is designated the *M period*. The times spent in each phase of mitosis are quite different. Prophase usually requires far longer than the other phases; metaphase is the shortest. DNA replication occurs before mitosis in what is termed the *S* (synthesis) *phase* (Fig. 1-4). In nucleated cells, DNA synthesis starts at several positions on each chromosome, thereby reducing the time required to replicate the sister chromatids. The period between M and S is designated the G_2 *phase* (post-DNA synthesis). A long G_1 *phase* (pre-DNA synthesis) follows mitosis and precedes chromosomal replication. Interphase includes G_1, S, and G_2. The four phases (M, G_1, S, G_2) constitute the life cycle of a somatic cell. The lengths of these phases vary considerably from one cell type to another. Normal mammalian cells growing in tissue culture usually require 18 to 24 hours at 37°C to complete the cell cycle.

2. Meiosis.

Sexual reproduction involves the manufacture of gametes (*gametogenesis*) and their union (*fertilization*). Gametogenesis occurs only in specialized cells (*germ line*) of the reproductive organs. Gametes contain the haploid (n) number of chromosomes, but originate from diploid ($2n$) cells of the germ line. Obviously the number of chromosomes must be reduced by half during gametogenesis. The reductional process is called *meiosis* (Fig. 1-5). Meiosis actually involves two divisions. The first meiotic division (meiosis I) is a reductional division producing two haploid cells from a single diploid cell. The second meiotic division (meiosis II) is an equational division which separates the sister chromatids of the haploid cells. The prophase of meiosis I differs from the prophase of a mitotic division in that homologous chromosomes come to lie side by side in a pairing process called *synapsis*. Each pair of synapsed homologues is called a *bivalent*; since it consists of four chromid strands, a bivalent is also called a *tetrad*. During synapsis non-sister chromatids may break and reunite with one another in a process called *crossing over*. The point of exchange

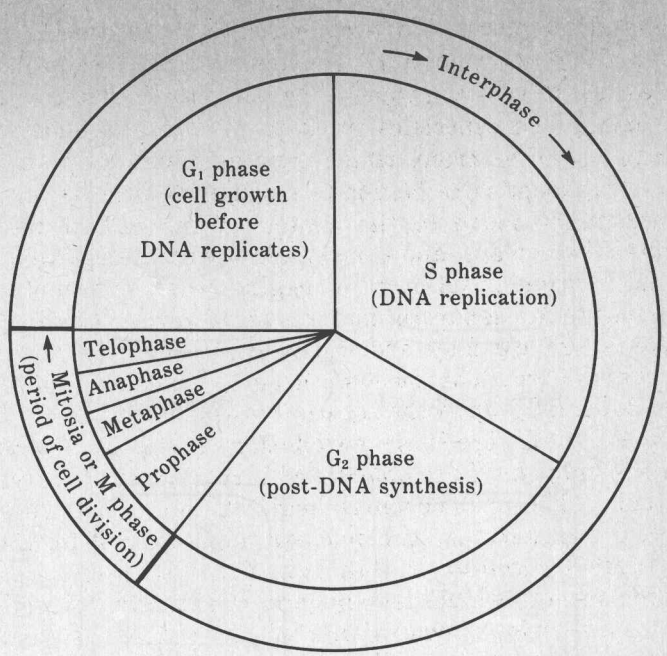

Fig. 1-4. Diagram of a typical cell reproductive cycle. (Idea from Jack A. Ward & Howard R. Hetzel, *Biology: Today and Tomorrow*, p. 71, 1980, West Pub.)

appears in the microscope as an overlapping region called a *chiasma* (chiasmata, plural). During metaphase I, the bivalents orient themselves at random on the equatorial plane. At first anaphase the centromeres do not divide, but continue to hold sister chromatids together. The homologues separate and move to opposite poles. That is, whole chromosomes (each consisting of two chromatids) move apart. This in effect is the movement which reduces the chromosome number from the diploid ($2n$) condition to the haploid (n) state. Cytokinesis in telophase I divides the diploid mother cell into two haploid daughter cells. This ends the first meiotic division. The brief period between the first and second meiotic divisions is called *interkinesis*. In prophase II, the spindle apparatus reforms. By metaphase II, the centromeres have lined up on the equatorial plane. During anaphase II the centromeres of each chromosome divide, allowing sister chromatids to separate. Cytokinesis in telophase II divides the two cells into four meiotic products.

The distinguishing characteristics of mitosis and meiosis are summarized in Table 1.2 below.

MENDEL'S LAWS

Gregor Mendel published the results of his genetic studies on the garden pea in 1866 and thereby laid the foundation of modern genetics. In this paper Mendel proposed some basic genetic principles. One of these is known as the *principle of segregation*. He found that from any one parent, only one allelic form of a gene is transmitted through a gamete to the offspring. For example, a plant which had a factor (or gene) for round shaped seed and also an allele for wrinkled shaped seed would transmit only one of these two alleles through a gamete to its offspring. Mendel knew nothing of chromosomes or meiosis, as they had not yet been discovered. We now know that the physical basis for this principle is in first meiotic anaphase where homologous chromosomes segregate or separate from each other. If the gene for round seed is on one chromosome and its allelic form for wrinkled

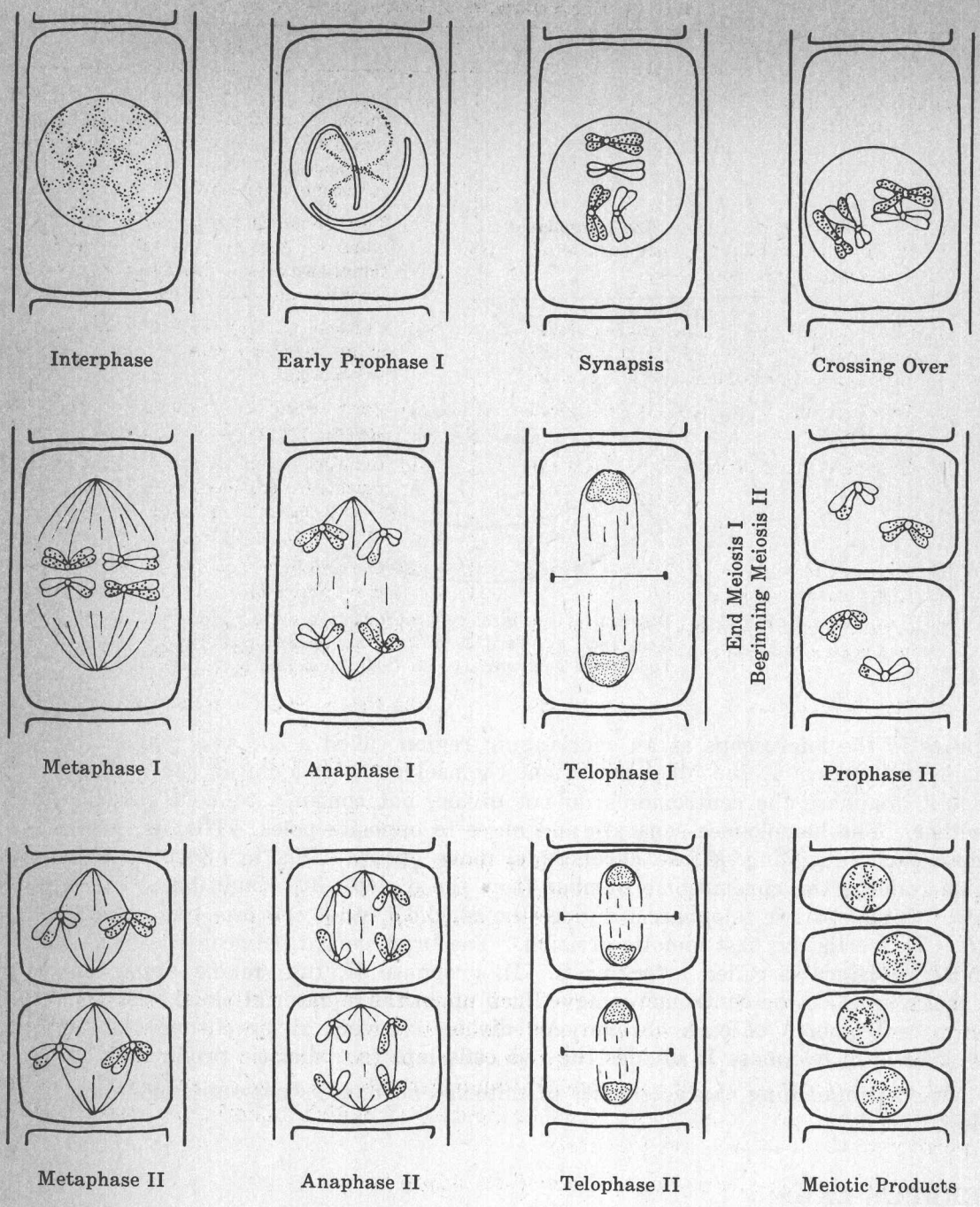

Fig. 1-5. Meiosis in plant cells.

seed is on the homologous chromosome, then it becomes clear that alleles normally will not be found in the same gamete.

Mendel's *principle of independent assortment* states that the segregation of one factor pair occurs independently of any other factor pair. We know that this is true only for loci on non-homologous chromosomes. For example, on one homologous pair of chromosomes are the seed shape alleles and on another pair of homologues are the alleles for green and yellow seed color. The segregation of the seed shape alleles occurs independently of the segregation of the seed color alleles because each pair of homologues behaves as an independent

Table 1.2. Characteristics of Mitosis and Meiosis

Mitosis	*Meiosis*
(1) An equational division which separates sister chromatids.	(1) The first stage is a reductional division which separates homologous chromosomes at first anaphase; sister chromatids separate in an equational division at second anaphase.
(2) One division per cycle, i.e. one cytoplasmic division (cytokinesis) per equational chromosomal division.	(2) Two divisions per cycle, i.e. two cytoplasmic divisions, one following reductional chromosomal division and one following equational chromosomal division.
(3) Chromosomes fail to synapse; no chiasmata form; genetic exchange between homologous chromosomes does not occur.	(3) Chromosomes synapse and form chiasmata; genetic exchange occurs between homologues.
(4) Two products (daughter cells) produced per cycle.	(4) Four cellular products (gametes or spores) produced per cycle.
(5) Genetic content of mitotic products are identical.	(5) Genetic content of meiotic products different; centromeres may be replicas of either maternal or paternal centromeres in varying combinations.
(6) Chromosome number of daughter cells is the same as that of mother cell.	(6) Chromosome number of meiotic products is half that of the mother cell.
(7) Mitotic products are usually capable of undergoing additional mitotic divisions.	(7) Meiotic products cannot undergo another meiotic division although they may undergo mitotic division.
(8) Normally occurs in most all somatic cells.	(8) Occurs only in specialized cells of the germ line.
(9) Begins at the zygote stage and continues through the life of the organism.	(9) Occurs only after a higher organism has begun to mature; occurs in the zygote of many algae and fungi.

unit during meiosis. Furthermore, because the orientation of bivalents on the first meiotic metaphase plate is completely at random, four combinations of factors could be found in the meiotic products: (1) round-yellow, (2) wrinkled-green, (3) round-green, (4) wrinkled-yellow.

GAMETOGENESIS

Usually the immediate end products of meiosis are not fully developed gametes or spores. A period of *maturation* commonly follows meiosis. In plants, one or more mitotic divisions are required to produce reproductive spores, whereas in animals the meiotic products develop directly into gametes through growth and/or differentiation. The entire process of producing mature gametes or spores, of which meiotic division is the most important part, is called *gametogenesis*.

1. Animal Gametogenesis (*as represented in mammals*).

Gametogenesis in the male animal is called *spermatogenesis* (Fig. 1-6(*a*)). Mammalian spermatogenesis originates in the germinal epithelium of the seminiferous tubules of the male gonads (testes) from diploid primordial cells. These cells undergo repeated mitotic divisions to form a population of *spermatogonia*. By growth, a spermatogonium may differentiate into a diploid *primary spermatocyte* with the capacity to undergo meiosis. The first meiotic division occurs in these primary spermatocytes, producing haploid *secondary spermatocytes*. From these cells the second meiotic division produces four haploid meiotic products called *spermatids*. Almost the entire amount of cytoplasm then extrudes into a long whiplike tail during maturation and the cell becomes transformed into a mature male gamete called a *sperm cell* or *spermatozoan* (-zoa, plural).

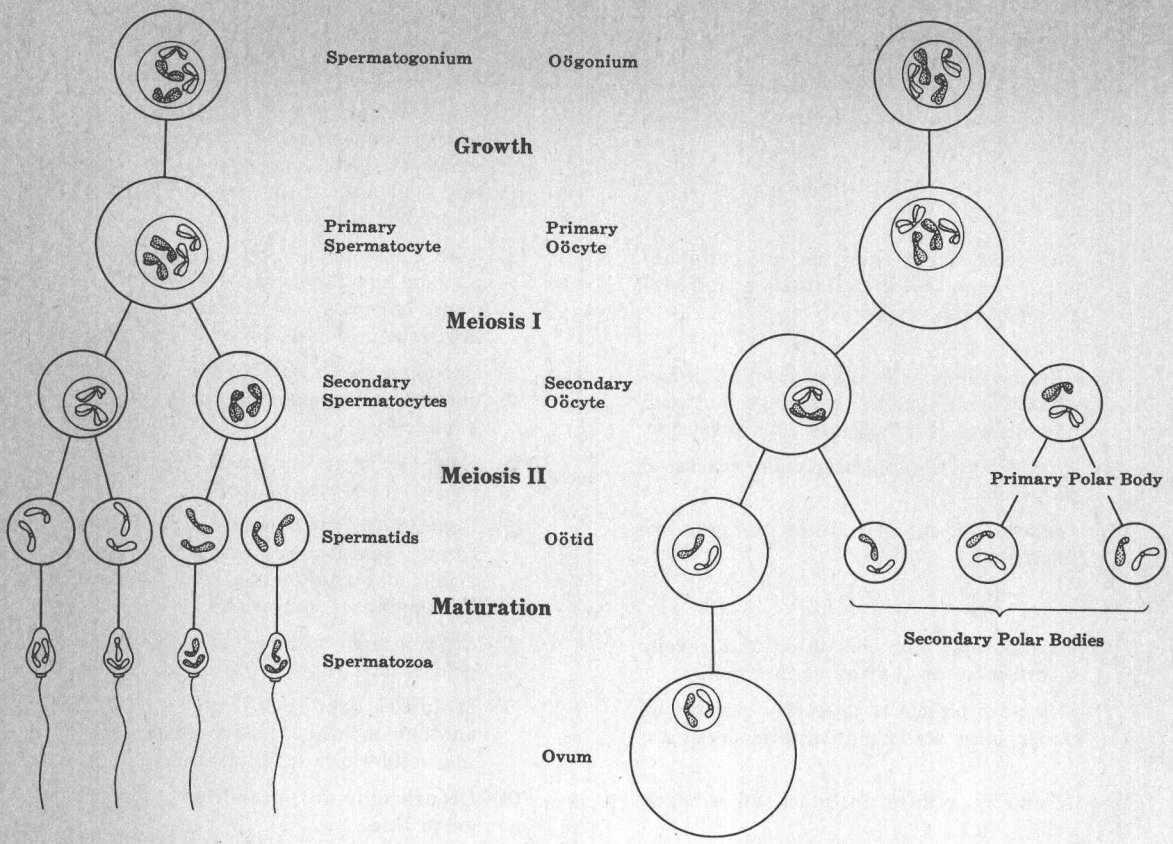

Fig. 1-6. Animal gametogenesis.

Gametogenesis in the female animal is called *oögenesis* (Fig. 1-6(*b*)). Mammalian oögenesis originates in the germinal epithelium of the female gonads (ovaries) in diploid primordial cells called *oögonia*. By growth and storage of much cytoplasm or yolk (to be used as food by the early embryo), the oögonium is transformed into a diploid *primary oöcyte* with the capacity to undergo meiosis. The first meiotic division reduces the chromosome number by half and also distributes vastly different amounts of cytoplasm to the two products by a grossly unequal cytokinesis. The larger cell thus produced is called a *secondary oöcyte* and the smaller is a primary *polar body*. In some cases the first polar body may undergo the second meiotic division, producing two secondary polar bodies. All polar bodies degenerate, however, and take no part in fertilization. The second meiotic division of the oöcyte again involves an unequal cytokinesis, producing a large yolky *oötid* and a secondary polar body. By additional growth and differentiation the oötid becomes a mature female gamete called an *ovum* or *egg cell*.

The union of male and female gametes (sperm and egg) is called *fertilization* and reestablishes the diploid number in the resulting cell called a *zygote*. The head of the sperm enters the egg, but the tail piece (the bulk of the cytoplasm of the male gamete) remains outside and degenerates. Subsequent mitotic divisions produce the numerous cells of the embryo which become organized into the tissues and organs of the new individual.

2. Plant Gametogenesis (*as represented in angiosperms*).

Gametogenesis in the plant kingdom varies considerably between major groups of plants. The process as described below is that typical of many flowering plants (*angio-*