Genetics of Domestic Animals

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PRENTICE HALL, Englewood Cliffs, New Jersey 07632

Stufflebeam, Charles E.

Cherrics of domestic animals Charles E. Stufflebram,
p. cm,
includes index.
ISSN 0-16-285124-2
1. Domestic animals--Denetics. 2. Animal breading. I. Title.
SF105.592-1989
886.08-21--9619
686.78-21

Editorial/production supervision: Merrill Peterson

Interior design: Joan L. Stone

Cover design: Diane Saxe

Manufacturing buyer: Bob Anderson



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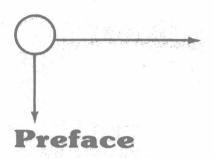
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Printed in the United States of America

10 9 8 7 6 5 4 3 2 1

ISBN 0-13-351214-2

Prentice-Hall International (UK) Limited, London
Prentice-Hall of Australia Pty. Limited, Sydney
Prentice-Hall Canada Inc., Toronto
Prentice-Hall Hispanoamericana, S.A., Mexico
Prentice-Hall of India Private Limited, New Delhi
Prentice-Hall of Japan, Inc., Tokyo
Simon & Schuster Asia Pte. Ltd., Singapore
Editora Prentice-Hall do Brasil, Ltda., Rio de Janeiro



The materials in this book have been designed for second and third year college and university students with an interest in domestic animals. It should be well suited for students in agriculture, biology, and veterinary medicine. Many excellent genetics textbooks are available, but very few of them use domestic animals as their primary models.

The book is designed so that only a minimal level of preparation in biology, chemistry, and mathematics is needed, most of which could be obtained at the secondary level or first year in college. A knowledge and understanding of the contents of this book should prepare a student very well for an advanced undergraduate or graduate course in genetics or animal breeding.

Each chapter begins with an outline of the material presented, which should be helpful in previewing the chapter, as well as serve as a guide for later review. Study questions, exercises, and problems follow each chapter to assist and guide students and instructors in the study of the principles and concepts presented. A list of other reading sources is also included at the end of each chapter.

The book is divided into three distinct sections. The first part deals with the biology of the cell and chemistry of the gene. The second part, chapters two through nine, covers qualitative genetics—basic principles of genetics. Topics included here are one and two pair crosses, multiple alleles, sex-related inheritance, linkage, epistasis, and probabilities. The third part deals with "quantitative genetics"—selection, inbreeding, and outbreeding.

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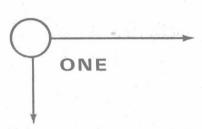
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Genetics is a biological science that deals with heredity and variation. Heredity involves the transmission of genetic material (genes) from one generation to the next. Variation includes the differences that can be seen among characteristics within each species. Differences among animal characteristics are brought about, in part, by the transmission of changed genes (mutations) or new combinations of existing genes. Environmental factors also play a significant role as a source of variation. The basic pattern of a trait or characteristic is determined by heredity; the development of the trait is then influenced in varying degrees by environment, as well as by other genes.

Genetics as a science began in the 1860s with the work of Gregor Mendel, an Austrian monk, who discovered the basic law of segregation and recombination of genes. He did not know about genes as such, but worked out the one-pair and two-pair ratios which are basic to the study of genetics. The results of Mendel's work with garden peas were published in 1865, but remained in relative obscurity for nearly 35 years. In 1900, three other scientists, working independently of each other, rediscovered Mendel's principles and then found the results of his work.

1.1 CELLS AND CHROMOSOMES

The smallest structural unit of an animal's body is the *cell* (Figure 1.1). A typical cell is microscopic, varying in size from approximately 10 to 20 micrometers in diameter (1 micrometer equals approximately $\frac{1}{25}$ inch). The

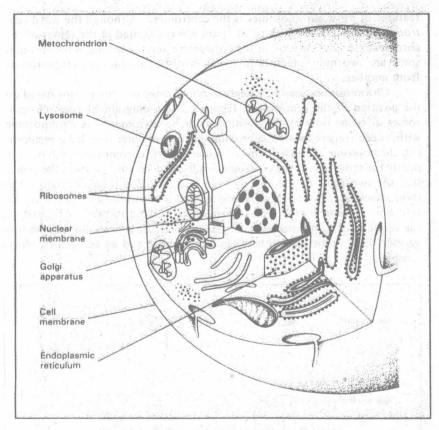


Figure 1.1 Sections and parts of a typical animal cell.

outer covering of an animal cell is the *cell membrane*. Inside the cell membrane are the *cytoplasm* and the *nucleus*. Contained within the cytoplasm are a number of *organelles* that are associated with the many vital functions of the cell. Included among these organelles are the mitochondria, the endoplasmic reticulum, lysosomes, ribosomes, and Golgi bodies. The nucleus is covered with a membrane inside of which are found the chromosomes and genes. It is with the genes and chromosomes that the subject of genetics is most concerned.

1.1.1 Characteristics of Chromosomes

Chromosomes are relatively slender, threadlike strands of material that contain the units of inheritance called genes. One of the prominent

features of most chromosomes is the centromere. Although the word centromere means "central part," it is not always located in the center of the chromosome. The position of the centromere and the length of the chromosome are two major factors involved in distinguishing one chromosome from another.

Chromosomes may be categorized into three or four groups based on the position of the centromere. Figure 1.2 is a diagram of four chromosomes differing only by the position of their centromeres. A chromosome with its centromere located approximately in the center is called a metacentric chromosome. The two arms of a metacentric chromosome will be very nearly the same size. If the centromere is located far enough from the center that the two arms of the chromosome are distinctly unequal in size, the term submetacentric is used. An acrocentric chromosome is one with the centromere located very near the end. If the centromere is located at the very end of a chromosome, the term telocentric is used. Sometimes it is very difficult to distinguish between a telocentric and an acrocentric chromosome.

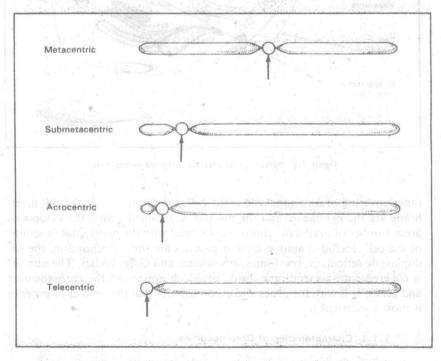


Figure 1.2 Four kinds of chromosomes categorized according to the positions of their centromères. (a portugal l'elles gone li gellai lo altinu esti mission tent

The overall length of chromosomes varies from a few to several micrometers. The number of genes may vary from a few on the very short chromosomes, to possibly thousands on the longer ones.

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The number of chromosomes in the nuclei of body cells is constant among animals of the same species. All body cells of cattle, for example, would be expected to have 60 chromosomes. If for some reason a zygote is formed that is missing one or more chromosomes, the genes that are found on those chromosomes will also be missing. Some of the missing genes could very well control functions so vital to the animal that it would not be able to survive without them. The presence of extra chromosomes results in the presence of extra genes, a situation that can cause metabolic problems for the animal. In fact, the chromosome number is so important that deviations from the normal affect the animal to the extent that it will usually die during embryonic development. The typical number of chromosomes in the body cells of several species is listed in Table 1.1.

In addition to the fact that there is a constant number of chromosomes in the body cells of animals, the chromosomes exist in pairs. The members of each of these pairs are of the same shape and length and have their centromeres located in the same position. The sex chromosomes represent an exception to this statement. The Y chromosome in male mammals is generally smaller than the X chromosome and does not carry as many genes. The situation regarding the Z and W chromosomes in female birds is similar. A more complete explanation of the characteristics and functions of the sex chromosomes is presented in Chapter Five.

Since the chromosomes in body cells do exist in pairs, it is customary to express chromosome number in terms of the number of pairs typical for the species, such as 19 pairs for swine, 39 pairs for dogs, and 23 pairs for humans. The two members of a pair are called *homologs* of each other. Homologous chromosomes are defined as two chromosomes that are alike

TABLE 1.1 Chromosome Numbers in Diploid Cells of Several Animal Species

The second secon	A Company of the Comp	and the second s	and halfer and the
Pig	38	Bison	60
Cat	38	Cattle	60
Mouse	40	Goat	60
Rat	42	Donkey	62
Rabbit	1010144 TOE	Horse	64
Human	46	Dog	78
Sheep	54	Chicken	78

in size, shape, and position of their centromeres. Homologous chromosomes may or may not contain some of the same genes.

Cells that contain pairs of chromosomes are described as being diploid. The symbol for diploidy is 2n. Body cells, also called somatic cells, are usually diploid. Sex cells, or gametes, are generally haploid (n). In gametes, the chromosomes are not paired. Only one chromosome from each of the homologous pairs is generally found in the nucleus of a gamete.

Figure 1.3 is a photograph of the chromosomes of a boar, taken during the metaphase of mitosis. The individual chromosomes have been cut out of the photograph and arranged by homologous pairs according to their overall length. The pairs of autosomes were then numbered from the largest to the smallest, with the sex chromosomes being placed last in the arrangement. Autosomies include all chromosomes except the sex chromosomes. Such an arrangement of chromosomes is called a *karyotype*. Karyotypes can be useful in determining chromosome number; they may be used to help



Figure 1.3 Photomicrograph of chromosomes of the pig arranged in a karyotype, with the chromosomes in pairs. At the metaphase of cell division when the chromosomes can be stained and observed, they appear doubled (sister chromatids) and are connected at the centomere. [From John F. Lasley, Genetics of Livestock Improvement, 4/E, © 1987, p. 19. Reprinted by permission of Prentice-Hall, Inc., Englewood Cliffs, NJ.]

determine the sex of embryos; and deviations from normal morphology and chromosome number can be identified by studying karyotypes.

1.1.3 The Concept of Alleles

The position or location on a chromosome where a particular gene is found is called a *locus*. Every gene has a locus. A pair of homologous chromosomes may or may not possess identical genes at the same locus. However, even when the genes at two homologous loci are not identical, they will affect the same trait. The different forms of the genes that can be found at the same loci on homologous chromosomes are called *alleles*. In summary, three key points characterize the definition of alleles: the genes are different; they occupy the same loci (positions) on homologous chromosomes; and they have their effects on the same trait. For example, a particular gene may cause a cow to develop horns, while its allele may cause another cow to be polled. The presence or absence of horns is the trait for which there are two alternate phenotypes or expressions: horns and polled. The term *phenotype* refers to the way in which genes express themselves.

When describing genes or solving genetics problems, it is necessary to use symbols to represent the genes. The most common practice is to use letters of the alphabet. There is no absolute standard to follow, but the most common way is to use upper- and lowercase letters to represent the alleles if only two are involved. Suppose that at one locus two alleles are possible. We can refer to this locus as the A-locus and let the letters A and a be the symbols for the two alleles. Either allele can occupy the locus on either homologous chromosome. In a group of animals it is very likely that all possible combinations of the two genes will exist. Some animals might possess two of the A genes, in which case the genotype would be written as AA. Other animals may possess two of the a genes and be designated as aa. Since each of these genotypes is made up of identical genes, they are referred to as homozygous (homo means "the same"). A third genotype is possible, the one that contains one of each of the two alleles (Aa). Since the genotype is made up of genes that are not identical, it is referred to as heterozygous (hetero means "different").

1.2 THE CHEMICAL NATURE OF GENES AND CHROMOSOMES

1.2.1 The Structure of DNA

A gene is composed of a substance known as deoxyribonucleic acid (DNA). DNA consists of two relatively long strands of material twisted to form a helix or spiral-like structure (Figure 1.4). The chromosomes are

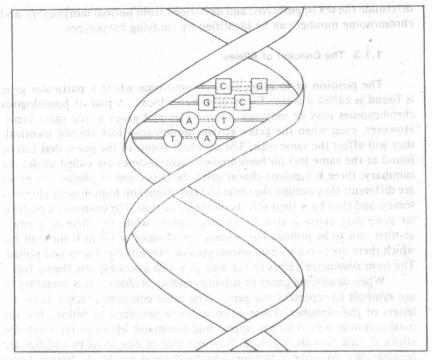


Figure 1.4 Diagrammatic representation of the Watson-Crick DNA structure. P, Phosphate; S, sugar; A, adenine; T, thymine; G, guanine; C, cytosine. The horizontal parallel lines symbolize hydrogen bonding between complementary bases.

made up of a complex substance called nucleoprotein, which consists of the DNA strands and a special kind of protein. Each of the two strands of DNA is composed of thousands of units called nucleotides. Each nucleotide is composed of a special nitrogenous base, the sugar deoxyribose, and phosphoric acid. The linkage in each strand of DNA is formed by a chemical connection between the sugar (deoxyribose) of one nucleotide and the phosphoric acid of the next, as illustrated in Figure 1.6.

The two strands of DNA are held together by relatively weak connections between the nitrogenous bases of one strand and those of the other strand. These relatively weak chemical connections are composed of hydrogen bonding. Four kinds of nitrogenous bases are found in DNA: adenine. thymine, cytosine, and guanine. The nature of the hydrogen bonding is such that adenine and thymine are attracted to each other, as are cytosine to guanine. This phenomenon is referred to as the base-pairing principle. (Figure 1.5)

Prior to cell division, actually, as a part of the entire process of cell division, each chromosome manufactures a new chromosome identical to itself. The first step in this duplication process involves the untwisting and separation of the two strands in the DNA helix. Each original strand then manufactures a new strand by attracting the appropriate nucleotides as determined by the base-pairing principle. This process is illustrated in Figure 1.6.

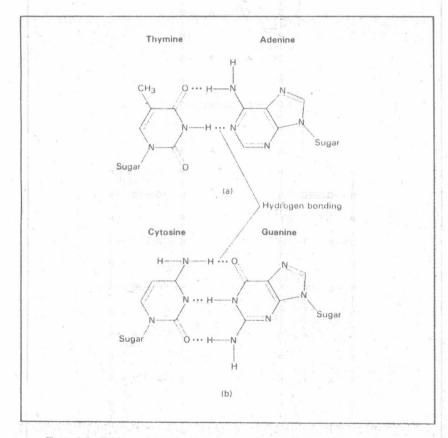


Figure 1.5 Pairings of rhymine and adenine (a), and cytosine and guanine (b), by means of hydrogen bonding as in DNA.

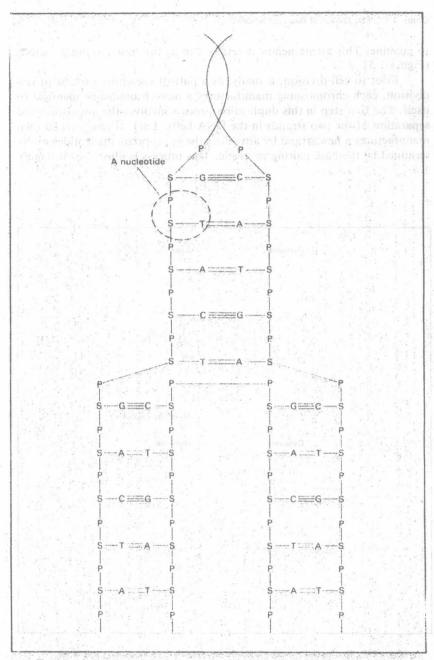


Figure 1.6 Illustration of a double-stranded DNA molecule duplicating itself. The two strands split and assemble the various parts on each strand, resulting in exact

1.2.2 Genetic Control of Protein Synthesis

One way that genes function is by controlling the production of enzymes that act as catalysts in various chemical reactions that take place in the cell, usually within the cytoplasm. Enzymes are proteins that are composed of amino acid subunits. Each enzyme is made up of a particular number and sequence of amino acids which is different from that of any other protein. The sequence of amino acids in each enzyme is determined by the sequence of nucleotides in the "coding" strand of DNA in the gene. A group of three nucleotides in the DNA is necessary to code a particular amino acid in the enzyme.

In order for the information carried in the genes to be transferred to the site of protein synthesis in the cytoplasm, a second kind of nucleic acid, called *ribonucleic acid* (RNA), comes into the picture. RNA is made up of the sugar ribose, phosphoric acid, and the four nitrogenous bases adenine, cytosine, guanine, and uracil. Uracil functions in RNA in much the same way as thymine does in DNA. In the process of base pairing, uracil pairs with thymine. The nucleotides in RNA are called *ribonculeotides*, while those of DNA are called *deoxyribonucleotides*. In summary, RNA differs from DNA in at least two ways; one is that RNA contains the sugar ribose instead of deoxyribose; the other is that RNA contains the base uracil in place of thymine.

One kind of RNA, called messenger RNA, or mRNA, is synthesized in the nucleus by the coding strand of DNA. One strand of DNA in a gene acts as a template in the synthesis of a molecule of messenger RNA. The mRNA, which consists of a single strand of nucleotides, contains the same number of nucleotides as are found in one strand of DNA in the gene from which it is patterned. Whenever the deoxyribonucleotides containing adenine, cytosine, guanine, and thymine appear in the template strand of DNA, the ribonucleotides containing uracil, guanine, cytosine, and adenine, respectively, are positioned in the molecule of mRNA. This is illustrated in Figure 1.7.

When the synthesis of a molecule of mRNA has been completed, it moves out of the nucleus and into the cytoplasm to form a complex with a ribosome. Ribosomes are relatively large structures usually associated with a system of membranes in the cytoplasm called the *endoplusmic reticulum*. The ribosomes are composed in part of ribonucleic acid, designated as *ribo*-

duplication of the molecule. P, phosphoric acid; S, sugar dioxyribose; A, base adenine; T, base thymine; G, base guanine; and C, base cytosine. Also note that a nucleotide containing P, S, and T is illustrated. [From John F. Lasley, Genetics of Livestock Improvement, 4/E, © 1987, p. 45. Reprinted by permission of Prentice-Hall, Inc., Englewood Cliffs, NJ.]