

# **Evolution of Sex Determining Mechanisms**

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**James J. Bull**  
University of Texas  
Austin, Texas



**1983**

**THE BENJAMIN/CUMMINGS PUBLISHING COMPANY, INC.**

Advanced Book Program  
Menlo Park, California

London • Amsterdam • Don Mills, Ontario • Sydney • Tokyo

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## Preface

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My interest in the evolution of sex determining mechanisms began when studying reptile chromosomes with R. J. Baker and G. A. Mengden during my undergraduate days at Texas Tech University. At this time Ohno's book *Sex Chromosomes and Sex-linked Genes* (1967) appeared. We were first of all inspired by Ohno's evolutionary perspective on sex chromosomes, and at the same time puzzled by the rigidly conserved systems of male heterogamety in mammals and female heterogamety in birds, compared with the existence of both systems in reptiles. Could male and female heterogamety be readily derived from an ancestor of either type? This question puzzled me for several years, and my association with two people, Richard Shine and Eric Charnov, helped me anticipate some possible solutions for this problem and thereby contemplate what has become the scope of this book. It was with R. Shine that I first considered the evolution of the sex ratio, which proved to be an important aspect in the evolution of sex determination. In this endeavor we both owe a major intellectual debt to G. C. Williams' *Adaptation and Natural Selection* (1966). Then with Eric Charnov at Utah, I began to directly address the topic of the evolution of sex determining mechanisms. Charnov taught me the basics of population genetics, while sharing with me his enthusiasm and insights into what he designated "sex allocation theory." Charnov was the inspiration for this book, having originally provided me with the confidence needed to accomplish my goals.

Since leaving Utah, I worked with several people who helped develop my understanding of theoretical population genetics: J. F. Crow, C. W. Cotterman, R. Lande, C. Denniston, K. Aoki, A. Gimelfarb, W. Engels, M. Moody, M. Rose (Wisconsin), J. Maynard Smith, B. and D. Charlesworth, P. Harvey (Sussex), and M. G. Bulmer (Oxford); and in my experimental work on turtles: J. M. Legler (Utah) and R. C. Vogt (Wisconsin). Charles Cotterman deserves special mention because of the inspiration he shared with me on the use and possibilities of the combinatorial method. Russell Lande also greatly influenced me by demonstrating how detailed genetic studies motivate population

genetics models of the evolutionary process. Raissa Berg offered valuable insights to the theory of genic balance.

This book was written from late 1981 to early 1983 while in residence at the University of Utah and was completed at the University of Texas. Drafts were read and criticized by Brian and Deborah Charlesworth, Paul Harvey, John Maynard Smith, Eric Charnov, and especially Sam Skinner (Utah). The first half of the book was read and criticized by David Crews and Allen Billy (U. Texas). H.-P. Bulnheim reviewed the chapter on cytoplasmic factors, K. Kallman reviewed the chapter on polyfactorial sex determination and other sections relevant to the fish literature, P. Anderson and B. Baker each reviewed the chapter on the genetic regulation of sex development, and J. Oliver provided a table of different genetic systems in mites and ticks. I thank all of them for useful suggestions and for correcting some of my misconceptions about the literature. Kerry Matz was delightfully creative and prompt in preparing the illustrations. Finally, Joni Moody provided her services toward editing the penultimate draft; only the few colleagues who read an early draft can appreciate the many improvements that stem directly from her work.

**James J. Bull**

## **PART ONE**

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# **Classical Sex Determining Mechanisms**

# 1

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## Introduction

### A. An Evolutionary Perspective of the Problem

This book is mostly about the diversity of sex determining mechanisms, or the manifold ways in which sex is inherited in animals with separate sexes. Beginning around 1900, the rediscovery of Mendelian genetics inspired studies which provided the first clear understanding of the inherited basis of males versus females in animals and plants, so that now, 80 years later, sex determining mechanisms have been evaluated in thousands of species. The demonstrated variety of sex determining mechanisms serves as an essential foundation for the evolutionary perspective proposed here, and I suggest ways in which sex determining mechanisms might further be studied to better understand the existing variety.

Evolutionary biology addresses the topic of variety among species, but there are two levels at which this variation is commonly investigated: the basic description of variety, and the processes which lead to the variety. Both levels of investigation will be adopted here.

#### The Classification of Variety: A Combinatorialist Perspective

In the simplest analysis one may observe that certain forms of a system are present and others are absent. This approach represents the combinatorialist

perspective, i.e., the categorization of known systems among those which are theoretically possible. The value of the combinatorialist approach is simply that it enables the investigator to consider the possible and perhaps probable existence of as yet undiscovered variety. This investigative process in turn may lead to the recognition of new variety or to the realization that some forms are invariably absent. To quote three inspiring combinatorialists,

We need scarcely add that the contemplation in natural science of a wider domain than the actual leads to a far better understanding of the actual.

(Eddington, 1929, pp. 266-7)

No practical biologist interested in sexual reproduction would be led to work out the detailed consequences experienced by organisms having three or more sexes; yet what else should he do if he wishes to understand why the sexes are, in fact, always two?

(Fisher, 1930, preface)

Often it will be found that the domain dreamed of by the combinatorialist is not really much wider than the actual.

(Cotterman, unpubl.)

One of the striking features of sex determination is the enormous variety of different types: male and female heterogamety, polyfactorial, environmental, and haplo-diploidy. And within these categories, there is further possible variety. For example, species with heterogametic sex determination may differ with respect to which sex is heterogametic, or with respect to the locus of heterogamety. Under environmental sex determination, sex is determined in response to temperature in some species but by other environmental factors in other species.

One of the major goals here will be to present the variety of mechanisms known and to cast this variety in terms of a larger set of possible mechanisms. For each of the major types of sex determination, the *known* mechanisms will be first described, followed by a presentation of the mechanisms *possible*. In some cases the alternatives are obvious and do not warrant enumeration. For example, under environmental sex determination, the possible variety of mechanisms is simply a list of the different environmental variables which could conceivably be used to determine sex. For other systems there is a well-defined set of possibilities, and a complete enumeration is desirable (e.g., male and female heterogamety).

The presentation of the variety of mechanisms known, coupled with the variety possible, could well stand on its own as a self-contained discipline in evolutionary biology. That is, one would be justified in looking no deeper. However, this appreciation for the variety of mechanisms is an almost essential prerequisite for the second objective, which is to understand how the different mechanisms evolve.

### The Processes which Generate Variety: Transitions

A second level of investigating variety is to consider how and why certain characteristics exist in populations—the processes that bring about change. The second objective, therefore, will be to describe the evolutionary transitions between different mechanisms. In some cases it is possible to describe *how* one mechanism changes to another; furthermore, it is sometimes possible to propose *why* such a change is favored. Heavy emphasis will be placed on descriptions of how sex determining mechanisms might change, especially for male and female heterogamety. Where empirical support of the transitions is lacking, some models will be offered as possibilities, not exclusive of others. For a few systems, such as environmental sex determination and haplo-diploidy, I will further suggest that there are general selective advantages for the transition. Based on this evolutionary framework, therefore, sex determining mechanisms will be presented along the following guidelines: (1) describe the variety known, (2) present the possible variety, (3) describe how the transitions might occur from one mechanism to another, and (4) consider why the transition might be favored.

Part I (Chapters 1-15) represents the major portion of the book and addresses almost exclusively the inherited and environmental bases of sex differences in animals. Under normal circumstances, sexual development is directed toward the production of a male or a female by an initial set of factors, such as the XX/XY genotype, and then proceeds as a highly integrated series of genetic and physiological steps which results in a male or female. These chapters will consider the genetic and environmental factors exerting the initial influence on which developmental pathway is to be followed, without considering the details of how the steps within each developmental pathway are coordinated. The chief reason for excluding a serious consideration of the details of sex development and differentiation is that the evolution of the inherited basis of sex may be described without understanding these various details. Rather, in its simplest form, an evolutionary model merely needs to specify inheritances and fitnesses of the different phenotypes, along with population breeding structure, all of which can be observed in ignorance of the physiological and molecular interactions which bring about these events. Chapter 4 does, however, review recent studies of genetic regulation of sex development in mammals, *Drosophila*, and *Caenorhabditis*.

Part II (Chapters 16-18) investigates the structural differences in the X and Y sex chromosomes. The evolution of sex chromosome differences is not to be equated with the evolution of sex determination, since the changes which occur on the sex chromosomes involve genes linked to those determining sex. However, the evolution of sex chromosome differences does indeed depend on the sex determining mechanism, and so sex chromosome evolution will be considered in this book as well.

Readers may wish to know how the present book relates to the recent literature on the evolution of sex. Four previous books (Williams, 1975; Maynard

Smith, 1978; Bell, 1982; Shields, 1982) were concerned with the evolution and maintenance of sexual reproduction, in which there are several major problems (Fig. 1.A). From a chronological perspective, the first steps in the evolution of sex may have been the evolution of biparental reproduction, or gametic union, followed by evolution of the male/female phenomenon, or small and large gamete producers, respectively. Superimposed upon these is the possibility for the evolution of parthenogenesis, in which biparental reproduction is abandoned. These problems rest completely outside the scope of this book. The topics addressed below concern sex determining mechanisms in populations maintaining sexual reproduction between males and females, while excluding questions on the origin of males and females or the evolution of parthenogenesis (Fig. 1.A).

The present book parallels and complements sex allocation theory (Charnov, 1982). Sex allocation theory considers the evolution of hermaphroditism (or sex change) versus separate sexes, the sex ratio, and the allocation to sperm versus eggs by a hermaphrodite. Sex allocation theory and the present book overlap with respect to the sex ratio: sex ratio selection appears to be the dominant force in molding sex determining mechanisms. However, these two bodies of theory diverge in the problems considered. This book will consider the evolution of mechanisms which produce the sex ratio, whereas sex allocation theory considers sex ratio evolution (or its counterpart in hermaphrodites) without regard to the sex determining mechanism.

## B. Terminology

The topic of sex, and its legitimate progeny, sex determination, is of certain interest to many people, with the result that various meanings may be implied by these terms until they are defined. Even among biologists, various meanings exist: sex may refer to gender, male or female, to the act of copulation, or to the process of gametic union (biparental reproduction as opposed to parthenogenesis). With respect to "sex determination" one again finds a term capable of encompassing multiple definitions, including the ascertainment of sex, the artificial manipulation of sex, or others. This problem with semantics has been recognized by several authors (McCarrey and Abbott, 1979; Austin et al., 1981), and in the effort to clarify the terminology used in this book, this section provides definitions for a few key words which might otherwise prove ambiguous.

(1) *Sex and Sex Determination*: Sex is defined as gender, male or female, with sex determination defined by extension as gender determination, the natural means by which a son or daughter is produced.

(2) *Sex Determining Mechanisms*: In the sense that differences in pea color might be studied from various perspectives such as (1) classical genetics

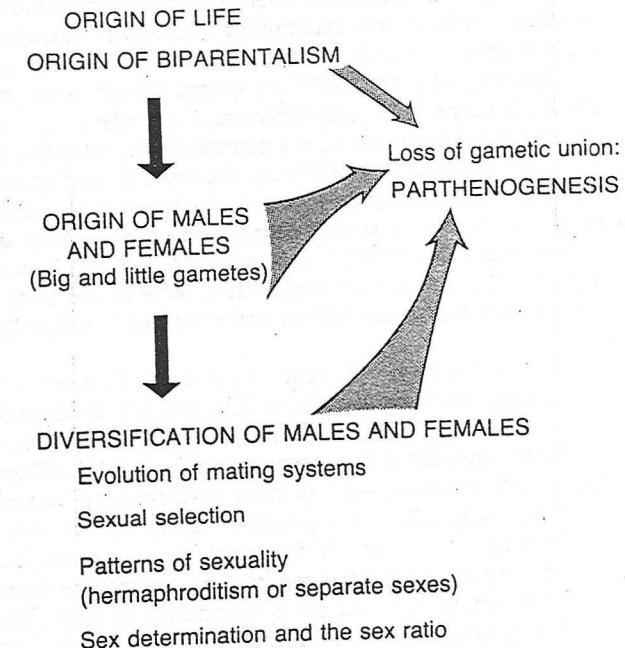


FIGURE 1.A. A perspective for the evolution of sexuality and related problems (based on the text of Maynard Smith, 1978). Historically, one of the first events in the evolution of sex may have been the development of biparental reproduction, in which offspring were produced from the union of two gametes at some stage of the life cycle. It is presumably difficult to know whether life itself originated with this form of reproduction, or if biparentalism was a subsequent development. Initially, biparentalism may have involved the union of equal-sized gametes (isogamy), as now found in some unicellular organisms. The next step in the evolution of sex would then have been a gamete dimorphism, so that small gametes always united with large ones (anisogamy), representing the essence of males and females. The origin of males and females then generated a new set of phenomena, such as sexual selection, the mating system, and sex determination. Superimposed on these processes is the possibility for the loss of biparental reproduction through the evolution of parthenogenesis. The present book is restricted to the topic listed at the bottom of the figure: sex determination and the sex ratio.

(inheritance of color), (2) molecular genetics (structure of color genes), and (3) physiology (biochemistry of pigment synthesis), a sex determining mechanism may also be described according to these different levels. Therefore, describing a mechanism as male heterogamety indicates the inherited basis of males versus females, but the mechanism may be equally described according to its physiology, molecular genetics, and so forth.

This book addresses the inherited and environmental basis of males versus females. The term "sex determining mechanism" therefore refers merely to a description at this level, without regard to other details such as the molecular or physiological properties of the system. It is sufficient that the reader understand this usage, but as reinforcement, I will refer to this level of description as the *classical* mechanism of sex determination, since this level of analysis has historical precedence. Specifically, the classical mechanism refers to the earliest elements in ontogeny common to one sex that distinguishes it from the other sex, including environmental and genetic effects acting in parents or zygotes that differentially influence the probability of producing or becoming male/female. For further clarification, some of the more common sex determining mechanisms are defined and explained in this context.

**Male Heterogamety**—All adult males are XY, adult females are XX. All zygotes are therefore XX or XY, and XY develops into male, XX into female. The X and Y are the inherited basis of sex determination, sex being determined at conception (in the zygote). (Chapter 2)

**Female Heterogamety**—As in male heterogamety, except that females are ZW, males ZZ. (The tradition of using the notation X and Y for male heterogamety, Z and W for female heterogamety, is retained here.) Depending on the meiotic mechanism, sex may be determined prior to or after fertilization, but sex is determined no later than the formation of the zygotic nucleus, so I shall refer to this as a zygotic mechanism. (Chapter 2)

**Polyfactorial (Polygenic) Sex Determination**—Sex is determined in the zygote by many factors with individually small effects, perhaps also with an arbitrary environmental effect. Thus the cumulative effect of many factors controls sex, rather than just two factors with a major effect. (Chapter 8)

**Environmental Sex Determination**—Sex is determined during embryogenesis in response to the local environment, with some environments producing males and others producing females. Sex cannot be predicted by zygotic genotype, because of the subsequent environmental influence. (Chapter 9)

**Arrhenotoky (Haplo-diploidy)**—A genetic system in which males arise from unfertilized eggs, females from fertilized eggs. Sex determination could be environmental (based on fertilization) or could be genotypic (based on ploidy or other). (Chapter 11)

(3) **Sex Development:** Although the emphasis in this book is on the classical sex determining mechanism, it is necessary to refer to other levels of sex determination, if only at a superficial level. To distinguish these other levels from the classical mechanism, the term "sex development" will be used. ("Sex differentiation" would be equally appropriate.) Sex development refers collectively to the various molecular, genetic, and physiological processes that produce a male or a female from a zygote of a given genotype and parents in a given environment.

(4) **Sex Factors:** Populations with separate sexes usually exhibit inherited differences in the probability of male or female determination. Consider male heterogamety: the inheritance of the X and Y can be said to determine sex because those zygotes inheriting two X's become female and XY becomes male. The segregating units that provide the inherited basis of differences in sex determination are defined as *sex factors*. In the simple systems of male and female heterogamety, the sex factors are X and Y, but multiple-factor systems have three or more factors, and polyfactorial systems may have several factors. It is also useful to distinguish between *major* and *minor* factors, the former having individually large effects and the latter small effects. Thus the X and Y of heterogametic systems are major factors, but the majority of sex factors in polyfactorial systems may be minor.

Sex factors may be defined as influencing sex determination either in the zygote or in the parents, controlling the offspring sex ratio according to the parental genotype. A *parental effect* on sex ratio would exist if, in the context of a particular mating, the average progeny sex ratio differed from that which would be obtained under Mendelian segregation of all sex factors from the parent, and sex determined among the zygotes according to their genotype and a random environment. This definition of parental influence excludes variation in family sex ratio due solely to differences in zygotic sex factors among the parents. Sex factors are assumed to be zygotic unless stated otherwise, and in this book there are in fact only a few mechanisms for which a parental effect is considered.

A sex factor may be inherited at a single locus or as several loci tightly linked on a chromosome (supergene). However, in the case of sex chromosomes, sex factors are inherited with many genes whose functions are independent of sex determination. I prefer to regard sex factors as the genes and their homologues responsible for controlling the inheritance of sex, and to differentiate them from genes which are incidentally co-inherited, even though in practice the sex factors may never be separated from the other linked genes. This definition, however, has the desirable effect of distinguishing the evolution of sex chromosome rearrangements and X-Y crossover suppression from changes in sex determination (the inheritance of sex). I consider these two processes as separate.

(5) **Sex Tendency:** A description of the inheritance of sex offers little information on how sex factors control sex development. For example, under male heterogamety, the X may dispose the embryo toward becoming female or the Y toward becoming male, or both, and the autosomes may also dispose the embryo toward one sex or the other. Beginning with Bridges (1922), the term "sex tendency" has been used to describe this underlying contribution toward male or female development. Specifically, a sex tendency is a number assigned to a gene (sex factor or otherwise) that reflects its contribution to whether the zygote develops as male or female, usually assumed to be invariant across different genotypes (see the additive-value model in Sec. 4.E). This concept was central to a model of gene action used by many early workers, often displayed

## PART I Classical Sex Determining Mechanisms

as  $M > F = \text{male}$ ,  $M < F = \text{female}$  (e.g., Witschi, 1929; Bridges, 1925; Winge, 1932, 1934, 1937; Hartmann, 1956; Bacci, 1965; Yamamoto, 1969). The M and F in these formulae refer to net male- and female-tendency values calculated for the entire genome.

"Sex tendency" is an attempt to describe the underlying mechanism of gene action in sex determination, and this concept is distinct from that of "sex factor" in three ways. First, one member of a sex factor pair may have no sex tendency even though the other member does, but this factor nonetheless influences the inheritance of sex by segregating opposite the one with sex tendency. Second, a gene may be assigned a sex tendency even if it does not segregate for differences and is therefore not a sex factor. Third, some mechanisms of sex determination are not compatible with this notion of sex tendency, and assigning values to sex factors does not lead to a valid description of sex determination. Most importantly, however, sex tendency is a *model* of gene action, which can be refuted by observation, whereas a sex factor is more an observation than a hypothesis.

(6) *Sex Chromosomes*: It is a common observation with male heterogamety (XX females/XY males) and female heterogamety (ZW females/ZZ males) that the X and Y (Z,W) are cytologically distinguishable from each other even though they pair at meiosis. The X and Y in this case are referred to as heteromorphic sex chromosomes, with the X carrying one sex factor and the Y the other. However, as described in Chapter 17, the sex factors are not always accompanied by cytological heteromorphism, as exemplified by some amphibians, fish and flies. To some readers, the use of the letters X and Y (or Z and W) may connote a cytological as well as genetic distinction. In Part I, a notational difference is used to distinguish cases in which X and Y are intended as cytological sex chromosomes from cases in which X and Y may be considered as the sex factors themselves, not tightly linked to other genes on the chromosome. Boldface letters (X, Y) are used when heteromorphic sex chromosomes are implied, lightface X and Y are used otherwise, when heteromorphism may apply but is not implied. Similarly, boldface A represents an autosomal chromosome (or haploid set of autosomes), whereas an italic A (as well as other italic characters) are used for an allele at a single locus.

J. L. A.

2

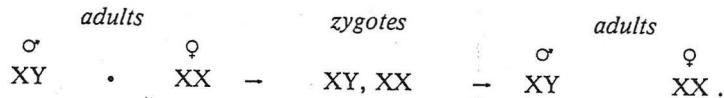
## Male and Female Heterogamety: 2-Factor Systems

The inheritance of sex may be influenced by three measurable effects: (i) major sex factors, (ii) minor sex factors, and (iii) environmental differences, and the study of sex determination is one of quantifying the relative magnitudes of these effects as well as their evolutionary consequences. This chapter and the following five chapters (2, 3-7) will consider mechanisms in which sex is inherited chiefly according to a few major factors. This chapter specifically considers XX/XY systems. A list is provided of taxa in which XX/XY systems are known, and tables are also provided for two properties of the X and Y which are not evident merely from their inheritance: the sex of YY, and recessive-X versus dominant-Y systems.

### A. Introduction

Male and female heterogamety are probably the two most common sex determining mechanisms known in animals, as they have been discovered in many invertebrates and vertebrates. Male heterogamety is described as an XX female/XY male system in which males produce X and Y sperm, females produce only X ova, and fertilizations yield either XX or XY zygotes. The system

is maintained because XX zygotes become female and XY zygotes become male:



This 2-factor system (the sex factors are X and Y) is denoted as *genotypic* sex determination because the X-Y genotype is the earliest ontogenetic characteristic distinguishing males from females. It is also a *zygotic mechanism*, because sex is determined in the zygote. Parents differing in their tendency to produce sons versus daughters, perhaps by altering the segregation ratios of X and Y, may offer a further variation of these mechanisms, and these will be designated as *parental mechanisms*. The X and Y *sex factors* may be inherited as a single locus, recombining freely with other genes on the chromosome, or they may be inherited in a tightly linked chromosome segment that incorporates other genes as well. Also, no assumptions are made concerning the molecular or physiological regulation of sex by the X and Y. These sex factors may consist of allelic differences at a single regulatory locus, or instead involve many tightly linked genes that collectively regulate sex. My definition of a sex factor does not address the nature of sex development, but rather concerns just the inheritance of sex.

Female heterogamety, denoted here as ZW female/ZZ male, differs from male heterogamety only in a reversal of the heterogametic sex. Both types of heterogamety are regarded here as zygotic mechanisms, even though in some cases of female heterogamety, meiosis may lead to the determination of sex prior to conception. A consequence of the formal similarity between male and female heterogamety is that arguments which apply to one system usually apply to the other. Henceforth, descriptions adopting the perspective of male heterogamety will be assumed to apply to female heterogamety unless indicated otherwise.

In some species, two or more *chromosomes* assort with respect to sex (e.g.,  $X_1X_1X_2X_2$  female/ $X_1X_2Y$  male, or XX female/ $XY_1Y_2$  male). These *multiple sex chromosome systems* (not to be confused with multiple-factor systems) are usually thought to have arisen by translocation of an autosome to the X or Y, as shown, or by fission of the X or Y (e.g., White, 1973):

	$\sigma$	$\varnothing$	Comments
Basic XX/XY system	AA	XX	AA XY
			Unlinked autosome indicated by A
Multiple-Y notation	$\ddot{XX}$	$\dot{X} \quad Y_1 \quad Y_2$	
interpretation	AA XX	A A Y $\dot{X}$	The neo-X ( $\dot{X}$ ) is a fusion of an autosome and the ancestral X

	$\sigma$	$\varnothing$	Comments
Multiple-X notation	$X_1X_1X_2X_2$	$X_1X_2\dot{Y}$	
interpretation	AA	XX	A X A $\dot{Y}$

The neo-Y ( $\dot{Y}$ ) is the ancestral Y fused to an autosome.

According to the definition of a classical sex determining mechanism, the evolution of a multiple sex chromosome system does not constitute a change in sex determination. The location of the sex factors does not change, but rather the sex linkage increases. There is in fact no fundamental difference in this respect between the evolution of multiple sex chromosomes and the evolution of crossover suppression along the XY chromosome pair. Therefore, the notation XX/XY includes multiple sex chromosome systems, except where indicated.

Other systems are known which lack a Y (XX/XO), or in which only the Y is present (OO/OY). Systems such as these are also regarded as two-factor systems because the regularity of meiosis ensures that the inheritance of sex is the same as with XX/XY. Indeed, even when X and Y chromosomes are both present, one almost never knows if the sex factor locus is present on both chromosomes.

### B. Diagnosing the Heterogametic Sex

There are commonly three methods of diagnosing the heterogametic sex in species with separate sexes: cytogenetics, sex linkage of markers, and the breeding of animals whose sex has been experimentally altered by environmental factors (reviewed in White, 1945, 1954, 1973; Westergaard, 1958; Gallien, 1959; Bacci, 1965; Crew, 1965; Mittwoch, 1967, 1973; Ohno, 1967).

#### Cytogenetics

The chromosomes of diploid animals generally occur in duplicate, with one copy from each parent. In meiosis, the chromosomes align together in pairs, and one member of every pair is transmitted to each gamete. Usually the two chromosomes from each pair, called homologues, are similar in size and shape. However, in many species, the study of chromosomes has revealed a consistent asymmetry involving one of the chromosome pairs in either the males or the females. This asymmetry is commonly manifested as an odd number of chromosomes, or as a heteromorphism—two homologues differing in size and/or shape. These heteromorphisms (or unpaired chromosomes) are usually the sex chromosomes (Fig. 2.B). One chromosome, the Y (W), is restricted to one sex, and its meiotic partner, the X (Z), occurs in both sexes,

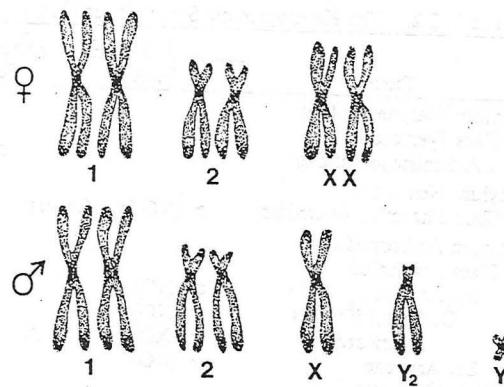


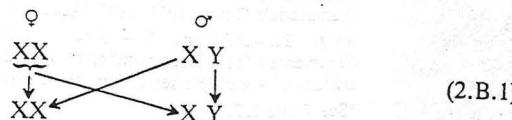
FIGURE 2.B. Sex Chromosome Heteromorphism. The diploid chromosome complement of the male and female muntjac deer is illustrated schematically. The chromosomes have been artificially arranged in homologous pairs, according to approximate size. The female has two X chromosomes, while the male has one X and two Y chromosomes (a condition known as multiple sex chromosomes). (Drawing based on D.H. Wurster and K. Benirschke, 1970. "Indian Muntjac, *Muntiacus muntjak*: a new deer with a low chromosome number." *Science* 168: 1364-6, Figure 1.)

although with multiple sex chromosomes, more than one X or Y are present. The majority of species in which the heterogametic sex has been reported have been studied by examination of chromosome heteromorphisms.

Cytological studies offer only indirect evidence for the presence of heterogametic sex determination, because chromosomal differences between the sexes could conceivably arise from reasons other than sex determination (e.g., sex-specific lethals). This objection is offered more in principle than in practice, as there do not appear to be many examples in which the cytological evidence of heteromorphic "sex" chromosomes has misidentified the sex factors. Further evidence that the X and Y chromosomes carry the sex factors may be obtained by observing the sex of (occasional) XYY or XO individuals (see Sec. 2.D). Of course, the absence of cytologically detectable sex chromosomes does not imply that heterogametic sex determination is lacking.

### Sex Linkage

The inheritance of X and Y sex factors is readily distinguished from autosomal inheritance:



Therefore, segregation of a visible marker which is sex-linked, or partly so, can be used to identify the heterogametic sex. Such polymorphisms have led to the identification of the heterogametic sex in a variety of animals: Y-linked color genes are known in a few fish and flies, Z-linked polymorphisms are known in some birds and lepidopterans, and many X-linked genes are known in flies and mammals (general reviews in Gallien, 1959; Bacci, 1965; Crew, 1965; for mammals, Ohno, 1967; fish, Atz, 1964; Yamamoto, 1969; Kallman, 1968; Lepidoptera, Cock, 1964; Johnson and Turner, 1979; flies, Tsukamoto et al., 1980; Franco et al., 1982). This method can reveal the heterogametic sex even if sex chromosomes are not cytologically distinguishable, but it requires the fortuity of detecting a polymorphism linked with the sex factor. It also suffers from some of the same limitations as the cytological approach in properly identifying the sex factors.

### Alteration of Sex by Environmental Factors

A more direct demonstration of heterogametic sex determination is possible if means are available to artificially circumvent genetic control of sex determination, such as with the external application of hormones, temperature, or other extrinsic factors. Suppose, for example, that hormones are used to produce males in all individuals. If the male is heterogametic, XX and XY males will be created, and mating of the XX males to normal (XX) females will produce all daughters. If the female is heterogametic, then some males will be ZW, and those mated to normal females will yield 1/4 ZZ, 1/2 ZW, and 1/4 WW. The sex ratio will depend on WW's sex and whether it survives. By producing unusual genotypes and sex ratios, this method diagnoses the heterogametic sex and provides information about how the X and Y influence sex determination. A variation of this method may be applied to species with occasional hermaphrodites. The hermaphrodite is bred to normal males and females, and the sex ratio is studied in both cohorts. Even though the sex genotype of the hermaphrodite is unknown, the alternatives of male and female heterogamety may be distinguished. A limitation of this method is that it assumes that sex is determined according to male or female heterogamety, and other sex determining mechanisms may provide results consistent with these expectations.

### C. Taxonomic Patterns

The following tables indicate many of the taxa in which male and female heterogamety are known, though these compilations are by no means complete (Tables 2.C.1-5). Some major groups are not represented here, but in some cases these groups consist primarily of species that are hermaphroditic, haplo-diploid, or have some other genetic system which does not facilitate the

evolution of sex chromosomes or heterogamety; the detail to which groups are listed reflects both the available information and the variety of heterogametic mechanisms within the group. Male heterogamety is extraordinarily prevalent in some of the invertebrates, such as nematodes, arachnids, and insects, but female heterogamety is not uncommon otherwise. It may be noted that the heterogametic sex is conserved in some groups, but that it varies within other groups, and the conserved system may be either male heterogamety (nematodes, arachnids, most insects, mammals) or female heterogamety (Lepidoptera and Trichoptera, birds).

The heterogametic sex is nearly always diagnosed from cytological evidence, i.e., the observation of a chromosomal difference between the sexes; but there are some cases in which the evidence is also genetic, based on observation of sex-linked markers or the sex ratios obtained from breeding "sex reversed" individuals. It is understood here that evidence of the heterogametic sex is purely cytological unless indicated otherwise. If instead the evidence is genetic, those cases are indicated by (G); if the evidence is both genetic and cytological, those cases are indicated by (B). Cytological evidence of the heterogametic sex is sometimes questionable, especially in fishes, so these tables should not be used as final sources on the heterogametic sex in any species of particular interest. Instead, individual entries in the table should be used as guides to the original sources (usually found in the reviews cited for each table).

TABLE 2.C.1. The Heterogametic Sex in Angiosperm Plants

Family	Heterogametic Sex	
	Male	Female
Moraceae	♂ (B)	
Polygonaceae	♂	
Caryophyllaceae	♂ (B)	
Amaranthaceae	♂ (G)	
Liliaceae	♂ (G)	
Cucurbitaceae	♂ (B)	
Caricaceae	♂ (G)	
Euphorbiaceae	♂ (G)	
Chenopodiaceae	♂ (G)	
Ranunculaceae	♂ (G)	
Vitaceae	♂ (G)	
Asteraceae		♀ (G)
Rosaceae		♀ (G)

Sources: Winge, 1931; Westergaard, 1958; Lloyd, 1974; Charlesworth and Charlesworth, 1978.

Note: Dioecy is uncommon in most of these families. (G)—indicates that the evidence of heterogamety is genetic; (B)—indicates that the evidence of heterogamety is both genetic and cytological; otherwise the evidence is cytological.

TABLE 2.C.2. The Heterogametic Sex of Invertebrate Groups

Taxon	Heterogametic Sex	
	Male	Female
Phylum Platyhelminthes		
Class Trematoda		♀
F. Schistosomatidae		
Phylum Nematoda		
Class Phasmida (4 Orders)	♂ (XO in 4 Orders)	
Phylum Arthropoda		
Class Arachnida		
Sc. Acarina	♂ (XO)	
O. Parasitiformes	♂ (XO)	
Acariformes	♂ (XO)	
Sc. Araneae	♂ (XO)	
Class Crustacea		
Sc. Branchiopoda	♂	♀ (B)
O. Anostraca	♂	
Sc. Ostracoda	♂ (some XO)	
Sc. Copepoda		♀
Sc. Malacostraca		
O. Decapoda	♂	
O. Isopoda	♂ (G)	♀ (B)
O. Amphipoda	♂ (G)	
Class Chilopoda	♂	
Class Insecta		
Sc. Apterygota	♂ (XO)	
Sc. Pterygota		
Div. Exopterygota		
O. Ephemeroptera	♂ (XO)	
Odonata	♂ (XO)	
Orthoptera	♂ (XO)	
Dermaptera	♂	
Isoptera	♂ (XO)	
Embioptera	♂ (XO)	
Plecoptera	♂ (XO)	
Psocoptera	♂ (XO)	
Hemiptera	♂ (XO)	
Div. Endopterygota		
O. Mecoptera	♂ (XO)	
Tricoptera		♀
Lepidoptera		♀ (B)
Diptera*	♂ (B)	♀ (B)
Siphonaptera	♂	
Neuroptera	♂	
Coleoptera	♂ (XO)	

Source: Data from White, 1973, Chs. 16, 17. More recent data substantially improve the information available for some groups: Grossman et al., 1981 (Trematoda); Triantaphyllou, 1973 and 1976; Chitwood and Chitwood, 1974 (Nematoda); Oliver, 1971, 1977 (Acarina).

Note: Sc.—Subclass; O.—Order; (XO)—a true XX/XO system is documented for a large number of species, the absence of Y not being an artifact of a multiple sex chromosome system.

\*See Table 2.C.3.

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TABLE 2.C.3. The Heterogametic Sex in Diptera

Family	Heterogametic Sex	
	Male	Female
Tipulidae	♂	
Culicidae	♂	
Chironomidae	♂	♀
Simuliidae	♂ (B)	
Tephritidae	♂	♀
Drosophilidae	♂ (B)	
Muscidae	♂ (B)	♀ (G)
Calliphoridae	♂	
Phoridae	♂ (B)	
Anthomyiidae	♂	

Source: Data from White, 1973, Chs. 16, 17. Chironomids and muscids are discussed further in Secs. 3.A, D, E.

TABLE 2.C.4. The Heterogametic Sex of Vertebrates

Taxon	Heterogametic Sex	
	Male	Female
Class Osteichthyes		
O. Anguilliformes		
F. Anguillidae		♀
Congridae		♀
O. Salmoniformes		
F. Bathylagidae	♂	
Galaxiidae	♂	
Salmonidae	♂ (B)	
Sternopychidae	♂	
O. Cypriniformes		
F. Cyprinidae	♂ (G)	
Parodontidae		♀
O. Siluriformes		
F. Bagridae		♀
Loricariidae	♂	
Siluridae	♂	
O. Myctophiformes		
F. Myctophidae	♂	
Neoscopelidae	♂	
Synodontidae		♀
O. Atheriniformes		
F. Goodeidae	♂	
Poeciliidae	♂ (G)	♀ (B)
Cyprinodontidae	♂ (B)	
O. Beryciformes		
F. Melampheidae	♂	

CHAPTER TWO Male and Female Heterogamety: 2-Factor Systems

TABLE 2.C.4. (continued)

Taxon	Heterogametic Sex	
	Male	Female
O. Gasterosteiformes		
F. Gasterosteidae	♂	♀
O. Scorpaeniformes		
F. Cottidae	♂	
O. Perciformes		
F. Belontiidae	♂	
Cichlidae	♂	
Gobiidae	♂	
Osphronemidae		♀
Scatophagidae	♂	
O. Pleuronectiformes		
F. Cynoglossidae	♂	
O. Tetraodontiformes		
F. Balistidae	♂	
Class Amphibia		
O. Anura (Frogs)		
F. Pipidae		♀ (G)
Ranidae	♂ (G)	♀
Leptodactylidae	♂	
Hylidae	♂ (G)	
O. Urodele (Salamanders)		
F. Salamandridae	♂	♀ (G)
Ambystomidae		♀ (B)
Proteidae		♀
Plethodontidae	♂	♀
Class Reptilia		
O. Testudines	♂	
O. Squamata*	♂	♀
Class Aves		♀ (B)
Class Mammalia	♂ (B)	

Sources: Fish: general reviews in Yamamoto, 1969; Ebeling and Chen, 1970; Gold, 1979; Sola et al., 1981; Additional observations on fish include Thorgaard, 1978; Johnstone et al., 1979 (Salmonidae); Yamamoto, 1975 (Cyprinidae); Moreira Filho et al., 1980 (Parodontidae); Murofushi et al., 1980 (Balistidae); Park and Kang, 1979; Park and Grimm, 1981 (Anguilliformes). No comprehensive reviews are available for the Amphibia, since most of the findings are recent; these references are the following: Mikamo and Witschi, 1964 (Pipidae); Richards and Nace, 1978; Schmid, 1980 (Ranidae); Iturra and Veloso, 1981 (Leptodactylidae); Kawamura and Nishioka, 1977 (Hylidae); Schmid et al., 1979; Gallien, 1959 (Salamandridae); Sessions, 1982; Humphrey, 1945 (Ambystomidae); Sessions, 1980 (Proteidae); Kezer and Sessions, 1979 (Plethodontidae). For the Reptilia, see Sec. 9.D. For birds, two major references are Ray-Chaudhuri, 1973; Tagaki and Sasaki, 1974. For mammals, see Sec. 4.C.

Note: Symbols and notation as in preceding tables.

\*See Table 2.C.5.

TABLE 2.C.5. The Heterogametic Sex in Lizards

Taxon	Heterogametic Sex	
	Male	Female
Infraorder Gekkota		
F. Gekkonidae		♀
Pygopodidae	♂	
Infraorder Iguania		
F. Iguanidae	♂	
Infraorder Scincomorpha		
F. Lacertidae		♀
Scincidae	♂	
Teiidae	♂	
Infraorder Platynota		
F. Varanidae		♀

Source: King, 1977. "The evolution of sex chromosomes in lizards." In: *Evolution and Reproduction*, edited by J. Calaby and H. Tyndale-Briscoe. Australian Academy of Science, Canberra, pp. 55-60, Table 1.

Note: F-Family.

In several species with heterogametic sex determination, the inheritance of sex may occasionally deviate from XX/XY, due either to environmental extremes or to rare combinations of autosomal sex factors (e.g., Secs. 8.B, 9.G). This variation may, on the one hand, provide the basis for the evolution of radically different sex determining mechanisms, and so it is important to acknowledge this phenomenon in the proper context (Secs. 8.F, 10.C). At the same time, occasional exceptions to an otherwise simple mechanism for the inheritance of sex do not warrant abandoning the overriding theme of simplicity. For the purposes of the combinatorial classification, a mechanism that determines sex according to XX females and XY males in 95% of the individuals may be classified as male heterogamety just as meaningfully as mechanisms in which sex is always inherited according to XX female/ XY male. Furthermore, there is no basis from an evolutionary perspective for distinguishing slight variations in heterogametic sex determination: models for the transition between different types of mechanisms seem robust to small magnitudes of "atypical" sex determination. Thus, without hesitation, I classify mechanisms as male and female heterogamety even if they are known to produce occasional exceptions.

#### D. Recessive-X and Dominant-Y Systems: The Sex of XO and XXY

The inheritance of sex under male and female heterogamety is described fully by XX/XY, but this formula provides only a rudimentary understanding of the nature of these sex factors. Heterogametic systems have sometimes been studied in special ways to further characterize the X and Y, and these studies

enrich our knowledge about the variety of systems that remains hidden when only the inheritance of sex is known. This section and the following one will consider two further aspects of heterogametic sex determination which have often been studied.

An XX/XY system may be accomplished by various possible mechanisms of gene action, as emphasized quite early by Muller (1932). Considering male heterogamety, for example, the X might have a female tendency or the Y a male tendency, or both, with the autosomes also exhibiting a sex tendency. Thus XX/XY may represent

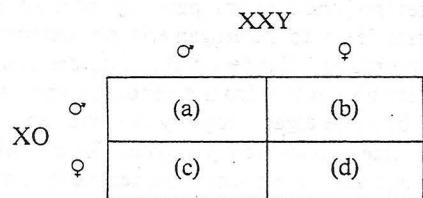
$$\begin{array}{ccc} & \text{♀} & \text{♂} \\ \text{or,} & \begin{array}{l} A_m A_m X_f X_f \\ A_f A_f X_o X_o \end{array} & \begin{array}{l} A_m A_m X_f Y_o \\ A_f A_f X_o Y_m \end{array} \\ \text{or,} & \begin{array}{l} A_o A_o X_f X_f \\ A_o A_o X_f Y_m \end{array} & \end{array}$$

or other combinations, where (A) represents a haploid set of autosomes, the subscript (o) the absence of sex tendency, (f) a female tendency, and (m) a male tendency (modified from Beermann, 1955; Bush, 1966). Sex could even depend upon heterozygosity versus homozygosity (XY versus XX), so that the concept of sex tendency was not even applicable (see Sec. 3.F).

Recognition of this possible variety within systems of male or female heterogamety has stimulated studies to identify the roles of X and Y in initiating sex development. One well-known approach to this problem has been to study the sex of individuals with an atypical complement of sex factors, such as XO and XXY. These studies have usually been conducted in species with heteromorphic sex chromosomes, so that the atypical individuals were distinguishable by their odd number of sex chromosomes. It is fortunate for this work that, in systems with heteromorphic sex chromosomes, the Y is often highly degenerate and does not seem to have nearly as many gene functions as the X. Consequently its presence or absence often does not affect viability.

Bridges (1916, 1925) was the first to use this approach, and he applied it to study sex determination in *Drosophila melanogaster*. This species normally has XX females and XY males, but through occasional accidents of meiosis, flies were obtained with a normal complement of autosomes but a sex chromosome constitution of XO or XXY. The former were males and the latter were females, as though the X played an active and recessive role in producing females, while the Y was passive, influencing sex merely by segregating opposite the X. In the study of these and other genotypes, Bridges further showed that sex was determined according to the number of X's relative to the number of autosomes, rather than according to the absolute number of X's: a fly with three sets of autosomes and two X's (3A + 2X) possessed some male characters, whereas a completely triploid 3A + 3X fly was fully female.

There have since been similar studies of a few other species (Table 2.D). If one considers whether XO and XXY individuals with two sets of autosomes are male or female (no intersexes), there are four possible outcomes:



However, species usually fall into one of two categories, which are denoted as follows:

**Recessive-X Systems** (b above): XO male, XXY female. Sex varies according to the presence of one or two X's, irrespective of the Y. (Previous terms include "genic balance" and "X-autosome ratio"; e.g., White, 1973, Ch. 16.)  
**Dominant-Y Systems** (c above): XO female, XXY male. Sex varies according to the presence of the Y, irrespective of the presence of one or two X's.

From Table 2.D, most species with a Y chromosome have a dominant-Y system, although a recessive-X system is known in a few cases. Of course, the many XX/XO species also have recessive-X systems (Table 2.C.2). Many studies infer dominant-Y and recessive-X systems from the analysis of atypical genotypes other than or in addition to these (e.g., polyploids), but these other genotypes are not included in this table.

The terms recessive-X and dominant-Y are convenient descriptions of these characteristics, and do not imply any particular model of gene action. One possible interpretation of both these systems is that one of the sex factors (X or Y) is essentially null regarding gene action in regulating sex development. Under the principle of sex tendency, this interpretation implies that only one member of a sex factor pair need have a prominent sex tendency, although it may be either the X or the Y. The autosomes may therefore also have a sex tendency. Consequently, there may be far more genes with sex tendency than there are sex factors, and apparently not all sex factors have a sex tendency. (Recall that "sex factor" is defined according to the inheritance of sex; "sex tendency" reflects a supposed underlying contribution toward developing as male or female and thus may apply to a locus even if there are no segregating differences.)

### E. The Sex of YY

The heterogametic mechanism may be further characterized as to whether or not YY is the same sex as XY. This characterization is again distinct from the

TABLE 2.D. The Sex of XXY and XO Individuals with a Normal Complement of Autosomes

Taxon	Sex of			Interpre- tation	References
	XY	XO	XXY		
<b>Mammals</b>					
<i>Homo sapiens</i> (man)	♂	♀	♂	Dom-Y	Cattanach, 1961; Ohno, 1967; Lyon et al., 1981
<i>Mus musculus</i> (mouse)	♂	♀	♂	Dom-Y	
<i>Bos taurus</i> (cow)	♂	—	I	?	Scott and Gregory, 1965
<i>Macropus eugenii</i> (wallaby)	♂	I(f)	I(m)	?	Sharman, 1970
<b>Diptera</b>					
<i>Drosophila melanogaster</i>	♂	♂*	♀	Rec-X	Bridges, 1925
<i>Musca domestica</i> (housefly)	♂	♀	♂	Dom-Y	Rubini et al., 1972
<i>Phormia regina</i> (blowfly)	♂	♀	♂	Dom-Y	Ullerich, 1963
<i>Lucilia cuprina</i> (blowfly)	♂	♀	—	Dom-Y	Ullerich, 1963
<i>Pales ferruginea</i> (tipulid)	♂	—	♂	Dom-Y	Ullerich et al., 1964
<i>Glossina palpalis</i> (tse tse)	♂	♂	♀	Rec-X	Southern, 1980
<b>Lepidoptera</b>					
<i>Bombyx mori</i> (silkworm)	♀	♂	♀	Dom-Y	Kihara, 1953
<b>Plants</b>					
<i>Melandrium (Silene) alba</i>	♂	—	♂	Dom-Y	Westergaard, 1958
<i>Rumex acetosa</i> group	♂	—	♀	Rec-X	Smith, 1963; Zuk, 1970a, 1970b

Note: I—Intersex, an individual with characteristics of both male and female; I(f)—intersex tending toward female; I(m)—intersex tending toward male.

This table excludes species which are regularly XX/XO, and excludes multiple sex chromosome systems (except for *Rumex*), since there is ambiguity as to which of the multiple X's or Y's carries the sex factor. A few of these references provide a more extensive listing of atypical sex chromosome genotypes, especially for *Rumex* (above references, plus Love, 1944), *Bombyx*, *Melandrium*, and *Drosophila*.

\*—XO is male in several other species of *Drosophila* (Voelker and Kojima, 1971; Hess and Meyer, 1968).

inheritance of sex, and the information is useful in revealing the nature of sex factors. Since YY does not normally occur in the population, some method is required to cause XY to develop as the same sex as XX; the mating of XY males to XY females then produces YY genotypes. There are two common methods of influencing the sex of XY: environmental effects (such as external application of hormones or temperature extremes), or additional sex factors (e.g., the W of the platyfish system generates WY females, so that YY can be obtained). The sex of YY is evaluated in the absence of these additional factors. A possible complication may ensue if YY is inviable, as commonly occurs in species with heteromorphic sex chromosomes (Chapter 16).

Table (2.E) lists cases in which the sex of YY is known. All examples conform to the rule that XY and YY are the same sex. The only clear exception to this rule is observed in a mechanism from some hymenopteran insects. These

TABLE 2.E. The Sex of YY Individuals

Taxon	Sex of			References
	XX	XY	YY	
<b>Fishes</b>				
<i>Oryzias latipes</i>	♀	♂	♂	Aida, 1936; Yamamoto, 1963
<i>Xiphophorus</i> (3 spp.)	♀	♂	♂	Kallman, 1983
<i>Poecilia reticulata</i>	♀	♂	♂	Winge and Ditlevson, 1947
<i>Carassius auratus</i>	♀	♂	♂	Yamamoto, 1975
<b>Amphibia</b>				
<i>Xenopus laevis</i> ZW	♂	♀	♀	Mikamo and Witschi, 1963, 1964
<i>Pleurodèles waltlii</i>	♂	♀	♀	Gallien, 1959; Collenot, 1975
<i>Ambystoma mexicanum</i>	♂	♀	♀	Humphrey, 1945, 1948
<i>Hyla arborea</i>	♀	♂	♂	Kawamura and Nishioka, 1977
<b>Diptera</b>				
<i>Musca domestica</i> *	♀	♂	♂	Milani et al., 1967; McDonald, 1971; Franco et al., 1982
<b>Crustacea</b>				
<i>Orchestia gammarella</i>	♀	♂	♂	Ginsburger-Vogel and Magniette-Mergault, 1981a,b
<i>Armadillidium vulgare</i>	♂	♀	♀	Juchault and Legrand, 1972
<b>Plants</b>				
<i>Thalictrum fendleri</i>	♀	♂	♂	Westergaard, 1958
<i>Asparagus officinale</i>	♀	♂	♂	Westergaard, 1958
<i>Mercurialis annua</i>	♀	♂	♂(st)	Westergaard, 1958

Note: X and Y are not necessarily to be interpreted as heteromorphic sex chromosomes. Instead, they may be inherited as single genes on a freely recombining pair of chromosomes. ♂(st)—a nearly sterile male.

\*—Applies both for a heteromorphic XY pair and also for cytologically indistinct factors.

hymenopterans have halpo-diploid sex determination, rather than male or female heterogamety, but the genetic basis of sex determination is such that individuals heterozygous for sex factors are female, and homozygotes (and haploids) are male (Sects. 3.F, 11.B). If these hymenopterans had heterogametic sex determination, XX and YY would develop as the same sex.

### F. The Origins of Heterogametic Sex Determination

In admiring the immense number of species with male or female heterogamety, it is natural to inquire how these systems arose. One might first entertain the possibility that, for some groups, heterogametic sex determination originated when separate sexes first arose. Of course, for some groups the origin of separate sexes is too remote for inferences about the sex determining mechanism at that time, but in other groups the origin of separate sexes is

quite recent, and one may study sex determination in these. For example, in flowering plants hermaphroditism is the common form of sexual reproduction, and in at least a few cases the origin of separate sexes (dioecy) is so recent that it can be traced to particular species. Substantial empirical and theoretical evidence demonstrates that one common pathway for the evolution of dioecy from hermaphroditism leads to heterogametic sex determination (Westergaard, 1958; Charlesworth and Charlesworth, 1978; described in Sec. 7.C).

Separate sexes may also evolve from a condition known as "isogamy," a condition illustrated by some algae. Isogamy involves reproduction in which two gametes of equal size join to form a zygote. The evolution of males and females (small- and large-gamete producers, respectively) conceivably could be the result of a dominant or recessive gene with a major effect on gamete size, and the establishment of such genes would in turn effect heterogametic sex determination. If the system is to accurately represent separate sexes, gametes of the same size must also be prevented from fusing, which could be accomplished if the gamete-size factor was linked to a mating-type locus. There have been some interesting models proposed for various aspects of this process, but there is not yet a strong empirical foundation for these evolutionary pathways (Parker, Baker, and Smith, 1972; Bell, 1982; Maynard Smith, 1978; Parker, 1982).

It should be emphasized that the above cases consider the origin of heterogametic sex determination as coinciding with the evolution of separate sexes. Many, if not the majority of existing systems of male and female heterogamety probably did not arise from a hermaphroditic or isogamous ancestor. Rather, they probably evolved in ancestors with separate sexes. That is, separate sexes have been continually present but the sex determining mechanisms have changed. These latter processes will be a major focus of this book.

### G. Summary

This chapter introduced the most widely recognized systems of sex determination, male and female heterogamety. Two factors control the inheritance of sex in both systems, so that one sex is heterozygous or heterogametic for sex factors and the other is homozygous for one of these factors. (The notation XX female/XY male is reserved for male heterogamety, ZW female/ZZ male for female heterogamety.) Heterogametic sex determination has evolved on many occasions and in different groups, so it provides a major focus for studies in the evolution of sex determination. The ubiquity of these systems further implies that they may provide a common ancestral state for the evolution of alternative sex determining mechanisms. A second purpose of this chapter was to illustrate some of the variety known within these mechanisms, regarding recessive-X and dominant-Y systems, and the sex of YY. This appreciation of variety provides a useful perspective for the introduction of the next chapter.