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# Chapter 1
# Extinct and Extant Reptiles: A Model System for the Study of Sex Chromosome Evolution

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**Abstract** The evolution and functional dynamics of sex chromosomes are focuses of current biological research. Although common organismal morphologies and functions of males and females are found among amniotes, underlying sex chromosome organizations and sex-determining mechanisms are widely variable. This chapter investigates the role that reptiles play in the study of sex chromosome evolution. Reptile studies have described the coevolution of genotypic sex determination and viviparity, the adaptive significance of sex-determining mechanisms, and shared ancestry of chromosomes. Novel resources, including whole-genome sequences and mapped sex-linked markers, have allowed researchers to examine sex chromosome evolution in reptiles, an important group for this type of study for their position as the sister group to mammals. Compared with mammals, reptiles exhibit much more variability in sex chromosome organization, providing raw material for study of sex chromosome evolution across amniotes.

## 1.1 Introduction

Embryos develop as either male or female depending on factors that vary widely among amniotes. Broadly speaking, amniotes can be classified as either genotypically sex-determined (GSD) or temperature-dependently sex-determined (TSD). Embryos of GSD species, including all mammals, birds, snakes, and many lizards and turtles, develop as either male or female depending on chromosomal contributions from parents at conception. Many, but not all, of these species exhibit detectable

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cytogenetic sex differences (i.e., heteromorphic sex chromosomes). The difference between heteromorphic and homomorphic sex chromosomes could be explained by the length of the interval since the origin of genotypic sex determination in a species (Ohno 1967; Janes et al. 2010b). Apparently, sex chromosomes begin to diverge from each other only after a new GSD system arises (see Sect. 1.3.1). This sex difference in karyotype is not apparent in individuals of TSD amniotes that develop as male or female primarily in response to incubation temperature, including all crocodilians, tuataras, and some turtles and lizards.

In this review, I will describe the variability of sex-determining mechanisms among amniotes. This variability includes, for example, the temperatures that trigger male or female development and the timing of temperature's effect among TSD species, as well as the presence or absence and type of sex chromosomes in GSD species. Almost all mammals exhibit male heterogamety in which females carry two X sex chromosomes of the same size and content, whereas males carry one X sex chromosome and one smaller, degenerated Y sex chromosome. In birds, females are heterogametic which means they carry the smaller, degenerated W sex chromosome and one larger, more gene-rich Z sex chromosome, whereas male birds carry two Z sex chromosomes. This difference in heterogamety affects the genomics of amniotes in ways that are discernible from genome sequencing and experimental evidence. Further, the evolutionary history of sex-determining mechanisms informs the different arrangements of amniotic sex chromosomes that have been studied using techniques that include phylogenetic inference, cytogenetic mapping, and measurements of population genetics parameters. Recent studies of sex-determining mechanisms and, specifically, the evolution of sex chromosomes have focused on extinct and extant reptiles for two reasons. First, nonavian reptiles exhibit greater variety of sex-determining mechanisms and sex chromosomes than birds or mammals. Second, genomic resources for reptiles (including birds) have recently improved to an extent that previously untestable hypotheses are now open to experimentation and comparative analyses (Janes et al. 2008).

## 1.2 Sex-Determining Mechanisms

### *1.2.1 Patterns and Variability*

Amniote sex-determining mechanisms are typically described as either GSD or TSD but within those categories, functional patterns vary. As described above, GSD species vary in their organization of sex chromosomes [i.e., female heterogamety (ZW system) or male heterogamety (XY system)] (Fig. 1.1a). Phylogenetic inference and comparative chromosome hybridizations suggest that male and female heterogamety have evolved more than once among amniotes although the exact number of independent origins is debated (Ezaz et al. 2009; Organ and Janes 2008). Likewise, the number of independent origins of temperature-dependent sex

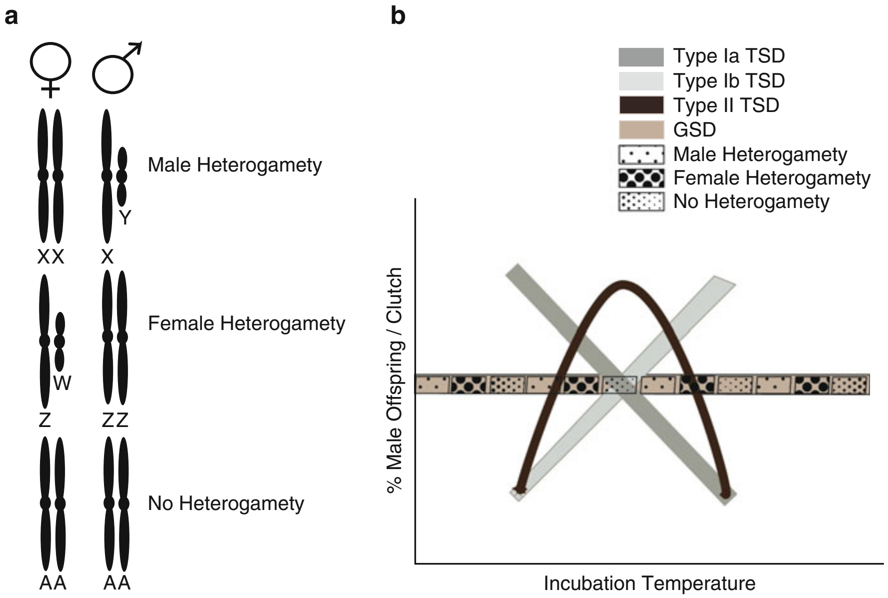

**Fig. 1.1** (**a**) Pairs of sex chromosomes that consist of either a male-specific Y chromosome and an X chromosome or a female-specific W chromosome and a Z chromosome. Species that exhibit these sex chromosomes are described as either male heterogametic (XY system) or female heterogametic (ZW system). Other GSD species exhibit no detectable heterogameties or sex differences in karyotype. (**b**) Influence of incubation temperature on offspring sex ratios among temperature-dependently (TSD) and genotypically sex-determined (GSD) species. The *y*-axis models the proportion of males yielded per clutch of eggs incubated at different points on the thermal gradient indicated on the *x*-axis. Sex-determining response to incubation temperature follows one of three patterns (Type Ia, Ib, or II) in TSD species. GSD species produce similarly balanced offspring sex ratios regardless of incubation temperature or type of heterogamety

determination is not clear. Although the sex-determining mechanisms of two or more species may respond to incubation temperature in a similar manner, the similarity may represent convergence. Three basic patterns of sex-determining response to incubation temperature (Types Ia, Ib, and II) have been described (Fig. 1.1b) (Bull 1983). Species that exhibit Type Ia temperature-dependent sex determination, such as loggerhead (*Caretta caretta*), green (*Chelonia mydas*), and leatherback (*Dermochelys coriacea*) sea turtles, produce more male offspring from eggs incubated at cooler temperatures (Standora and Spotila 1985). Species with Type Ib temperature-dependent sex determination, such as all crocodilians, produce more male offspring from eggs incubated at warmer temperatures (Valenzuela 2004). Species with Type II temperature-dependent sex determination, such as leopard geckos (*Eublepharis macularius*), produce a maximal proportion of males from eggs incubated at an intermediate temperature, whereas cooler or warmer temperatures yield higher proportions of females (Janes and Wayne 2006; Viets et al. 1994).

The timing of the effect of temperature on sex-determining response also varies among TSD reptiles. Shine et al. (2007) tested two TSD lizards for the effects of fadrozole, a chemical that blocks the bioconversion of testosterone to estrogen, thereby causing male development in eggs incubated at female-producing temperatures. In this type of experiment, the stage during which fadrozole affects offspring sex ratios represents the thermally sensitive period when temperature can influence sex determination. In two TSD reptiles, jacky dragons (*Amphibolurus muricatus*) and Duperrey's window-eyed skinks (*Bassiana duperreyi*), the thermally sensitive period in which sex could be reversed by fadrozole treatment occurred in the first half of the postoviposition incubation period. The thermally sensitive period has been shown to occur slightly later in turtles and tuataras, during only the middle third of the postoviposition incubation period (Ewert et al. 2004; Mitchell et al. 2006) and occurs even later in crocodilians, during the third quarter of the entire incubatory period (Lang and Andrews 1994).

GSD amniotes exhibit a similar degree of variability (Organ and Janes 2008). In birds, snakes, and some turtles and lizards, females are the heterogametic sex. Male heterogamety is found in some turtles and lizards and throughout mammals (with exceptions). The mammalian exceptions include, among others, the mole vole (*Ellobius lutescens*) in which a Y sex chromosome is absent. Both males and females of this species carry one X sex chromosome (Just et al. 1995; Vogel et al. 1998). Within heterogameties, there is variation in the extent of degeneration of either the male-specific Y sex chromosome or the female-specific W sex chromosome. For example, the Z and W sex chromosomes of emus (*Dromaius novaehollandiae*) are virtually homomorphic, whereas in chickens (*Gallus gallus*), the W sex chromosome is considerably smaller than the Z sex chromosome (Janes et al. 2009; Solari 1994). Clearly, a single line of demarcation between genotypic and temperature-dependent sex determination is overly simplistic and does not accurately represent the evolutionary history of sex-determining mechanisms in amniotes (Sarre et al. 2004).

### 1.2.2 Adaptive Significance of Sex-Determining Mechanisms

The variability of reptilian sex-determining mechanisms and, among GSD species, type of heterogamety are difficult to explain. Among agamid lizards, for example, species within the same genus with no discernible differences in natural history exhibit different sex-determining mechanisms (Ezaz et al. 2009; Uller et al. 2006). However, the adaptive significance of both genotypic and temperature-dependent sex determination has been explored in theory and experimentation. Fisher (1930) argued that parents should invest equally in sons and daughters. If sons and daughters represent equivalent parental investment, genotypic sex determination is expected to balance offspring sex ratios by matching them to the balanced probability of inheriting an X or a Y chromosome from a male parent in a male heterogametic species or the probability of inheriting a Z or a W chromosome from a female parent in a female heterogametic species. Charnov and Bull (1977) hypothesized that temperature-dependent sex determination would allow parents greater control over offspring sex ratios in environments where the costs of sons and daughters are unequal and fluctuating. However, the Charnov–Bull hypothesis has not acquired much empirical support. Parents of TSD species do not appear to control offspring sex ratios by nesting behavior. However, Freedberg and Wade (2001) suggested that offspring sex ratios are inherited as nest sites, and their unique exposures to sun and soil temperature are passed matrilineally. Also, Warner and Shine (2008) demonstrated that incubation temperature can affect reproductive success in jacky dragons. Male jacky dragons hatched from eggs incubated at the optimal male-producing temperature had greater lifetime reproductive success than males hatched from eggs incubated at a different temperature and experimentally masculinized by chemical aromatase inhibition. The same pattern of greater reproductive success was reported among females incubated at either the optimal female-producing temperature or a different temperature. This study provides evidence that, in a TSD species, incubation temperature directly influences reproductive success in a sex-differential manner. Although this study supports the Charnov–Bull hypothesis, it does not explain why some species would benefit from temperature-dependent sex determination but not other closely related species with similar life history traits.

Reproductive mode, whether a species is oviparous (egg-laying) or viviparous (live-bearing), is associated with type of sex-determining mechanism. Viviparity appears to be enabled by genotypic but not temperature-dependent sex determination. From a sample of 94 extant amniote species for which sex-determining mechanism, reproductive mode, and phylogenetic position are known, only two, perhaps three, exhibit both temperature-dependent sex determination and viviparity. The southern water skink (*Eulamprus tympanum*) and its sister species (*Eulamprus heatwolei*) give live birth and exhibit temperature-dependent sex determination and some evidence suggests that the spotted skink (*Niveoscincus ocellatus*) is also TSD and viviparous (Organ et al. 2009). For TSD species including these skinks, producing both male and female offspring requires exposing different embryos to one of at least two (optimal male-producing and optimal female-producing) thermal environments. For viviparous species, this requirement entails manipulating maternal body temperature and evidence for maternal manipulation of body temperature in TSD, viviparous skinks is debated (Allsop et al. 2006; While and Wapstra 2009). Further, as explained in Sect. 1.4, fluctuations in maternal body temperatures are even less likely in thermally consistent environments such as deep oceans. Apparently, thermal consistency is not an issue for oviparous, TSD species such as crocodilians and sea turtles because their nests experience sufficient thermal variation from top to bottom to explain mixed sex ratios emerging from clutches of eggs (Georges 1992 but see Warner and Shine 2009).