– ABSTRACT –

Among vertebrates, reptiles represent the ideal group for the study of sex determination. Reptiles include lineages with environmental sex determination (ESD) as seen in crocodiles and tuatara, lineages with genotypic sex determination (GSD), like e.g. iguanas, chameleons, skinks, lacertid lizards and birds, and few groups which possess variability in sex determination mechanisms, i.e. geckos, dragon lizards and turtles. This thesis is focused on the evolution of sex chromosomes and sex determination in turtles. The majority of turtle species exhibit ESD, which is considered the ancestral sex determination system of this group, while GSD either as male or female heterogamety evolved independently at least five times. We investigated the presence of sex chromosomes in representative species of turtles by cytogenetic analyses. The analyses included the reconstruction of karyotypes, distribution of constitutive heterochromatin (C-banding, methylation analysis) and repetitive elements (fluorescence *in situ* hybridization) and comparative genome hybridization (CGH), which often characterize the degenerated Y or W and can be helpful in the identification of "cryptic" sex chromosomes. We described XX/XY sex chromosomes in seven previously unstudied Australasian chelids (Pleurodira) from the genera *Chelodina*, *Elseya* and *Emydura*. Despite the difference in morphology, the accumulation of the same repetitive elements might suggest homology of male heterogamety in Chelidae and this family could represent another case of stable sex chromosomes, dating back to 50-120 million years ago. The presence of sex chromosomes was explored in three species of geoemydid and two species of emydid turtles. The previous report of heteromorphic ZZ/ZW sex chromosomes in the geoemydid *Pangshura smithii* was revealed to be based on an erroneous pairing of chromosomes during karyotype reconstruction, sex chromosomes were not detected in this species in our analyses. Notably, sex chromosomes were revealed neither in the geoemydid species *Geoemyda japonica* and *Geoemyda spengleri*, despite they are phylogenetically close to species with differentiated sex chromosomes, nor in the emydid taxa *Emys trinacris* and *Trachemys scripta scripta*. In addition, the evolution of sex chromosomes was explored in the first outgroup of turtles with GSD, birds. A qPCR-based method was applied to explore the homology of sex chromosomes across the bird radiation. The same method was also developed as a tool for molecular sex identification. In conclusion the main highlights from this thesis are that (i) male heterogamety is widespread and potentially old across Chelidae; (ii) the record of female heterogamety in Geoemydidae was questioned; as far as known, female heterogamety evolved only once in turtles in softshell turtles; (iii) the study of repetitive

elements is informative in the identification of cryptic sex chromosomes, (iv) sex determination is extremely conserved in birds and (v) a new qPCR-based molecular sex identification method, theoretically applicable in all bird species, was developed.