Sexual Development

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# Preface

The pathologies of abnormal sex determination and sex differentiation in vertebrates have been explored by integrated approaches at the interface of several disciplines. Disorders of sex development (DSD), a consensus terminology for the classification of ontogenetic error and pathological phenotypes associated with sex abnormalities recently adopted within the context of human medicine, has been extended to veterinary medicine for analogous pathologies of domestic animals. Detailed knowledge of the complex pathways extending from genetic sex determination to gonadal and phenotypic sex differentiation has arisen since Jost's [1947] postulate on the role of mammalian testicular function for masculinization of the corresponding male genitalia and the contrasting feminization of the genitalia and development of the female phenotype which occurs even in the absence of any differentiated gonad. The paramount discovery by Koopman et al. in 1991 that when introduced in the female genome, a specific region on the Y-chromosome (i.e. *SRY* gene) could drive the primitive bipotential gonad through the pathway of testicular determination, and eventually result in a normal male phenotype, further stimulated the scientific community to focus on the study of molecular and cellular processes involved in vertebrate sex determination and differentiation. The present themed issue on disorders of sex development in domestic animals aims to provide an updated review of information and scientific studies that have advanced our understanding of the occurrence of abnormal genetic, gonadal and phenotypic sex of livestock and some wild and captive exotic animals as well as present relevant current original research findings.

Radi and Parma open this compendium by reviewing the molecular mechanism of sexual development in mammals. The cascade of events that are triggered once the key transcription factor (e.g. *SRY* gene) is activated in the anlagen gonad are described, guiding the reader through the main cellular mechanisms that induce and regulate gonadal differentiation. Favetta et al. then provide a review article that presents examples of abnormal early development in association with DSD in domestic animals, emphasizing the cytogenetic and environmental factors which may be causatives of those pathologies. The well-known genetic goat XX sex-reversal syndrome is brought to our attention by Pannetier et al. and described in the light of the latest molecular and anatomopathological advances. Their review article exemplifies the level of complexity of study needed in order to understand a biological phenomenon as it is the lack of correspondence between the genetic sex and the phenotypic sex in mammals. Two subsequent broad review articles by Meyers-Wallen et al. and by Lear and McGee summarize the information available on gonadal and sex differentiation abnormalities of companion animal species and the domestic horse. Raudsepp et al. present an in-depth review focusing on the implication of the genomic organization of the sex chromosome pseudoautosomal regions on the occurrence of aneuploidies and concomitant sex disorders in domestic species. Mastromonaco et al. expand the information on DSD presenting data available for wild and captive exotic mammals. Closing the set of review articles of this themed issue, Lambert and Smith comprehensively provide an update of information on DSD in poultry.

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Seven original studies are included in this themed issue of *Sexual Development*. The article by Révay et al. documents the first mutation for the androgen receptor gene in domestic animals, which was found by DNA sequencing and proteomic studies of the androgen receptor in members of an equine family from the Quarter horse breed. This mutation offers the first evidence for an often predicted etiology for the well-known equine XY sex-reversal syndrome. Two other studies on sex disorders of the domestic horse, presented by Ciotola et al. and Das et al., add knowledge about the biological events that lead to ambiguous phenotypic sex presentation. Domestic dogs are also animals where several DSD have been previously described. Switonski et al. and Groppetti et al. describe the molecular and clinical findings of some individuals affected by DSD. Barasc et al. report the reproductive failure in the pig due to a Y-autosome translocation. Finally, Jeon et al. complement our compendium by presenting experimental data gained from investigating the levels of X-inactive specific transcript in somatic cell nuclear transfer embryos derived from female bovine freemartin donor cells.

Disorders of sex development in domestic animals was conceived as a themed issue intending to provide a comprehensive reference volume for those scientists, clinicians and students with interest in the field of sex ontogenesis of domestic vertebrates by providing an up-to-date base of knowledge for understanding abnormal sex development of animal species. We hope that the reader will enjoy the body of work presented in this exceptional issue.

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