

## **Abstract**

Sex development is a sophisticated regulatory system that is controlled by the activation and inhibition of specific genes. Gonadal differentiation is governed by the presence of the *SRY* gene on the Y chromosome. Expression of this gene triggers a cascade of events leading to the formation of testicular tissue; otherwise, in the absence of the Y chromosome and the *SRY* gene, the reproductive system develops towards the ovaries. As technologies in cytogenetics and molecular biology have advanced, diagnostic methods have improved and the molecular causes of DSD, which are both clinically and causally diverse, are being increasingly elucidated. There are several classification systems for DSD, and this thesis is based on the classification with three main diagnostic categories: 46,XX DSD; 46,XY DSD and sex chromosome anomalies. This system uses the patients' karyotype and histological characteristics of the gonads to identify and descriptively classify DSDs. This thesis briefly describes the categories of DSD, their characteristic features, and how they differ from each other. The main focus is on the categories 46,XX DSD and 46,XY DSD, previously referred to as sex reversals, which involves the development of phenotypic features that are contrary to the genotype of the individual. The 46,XX DSD category includes individuals with ovotesticular disorder of developmental, in which both testicular and ovarian tissue are present in the gonads. Moreover, it also includes individuals who have complete testicular development without any evidence of ovarian tissue. In most patients with 46,XY DSD, despite the presence of a Y chromosome, testicular tissue does not develop, and phenotypically, they appear as females. Finally, this thesis focuses on the category of sex chromosome aberration, which includes Turner's syndrome and Klinefelter's syndrome, two of the most common chromosomal aberrations affecting females and males. A common feature of all these categories is their high phenotypic variability, which represents a major obstacle in the diagnosis and subsequent treatment of these individuals.

**Keywords:** Disorders of sex development (DSD), sex reversale, ambiguous genitals, ovotestis, *SRY*