Genetic Steroid Disorders: Chapter 5. Androgen Insensitivity Syndrome

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After reviewing the mechanisms of androgen action, we will look at the main clinical features of androgen insensitivity, as well as the biological, cellular, and molecular tools used to investigate the AR. Numerous AR mutations have been described over the past 20 years. We will consider the challenges for diagnosis and prognosis by examining the genotype–phenotype relationship and the possibility of somatic mutations. Last, we will address the role of genetic counseling and the elements that should be taken into account for the difficult decision of sex assignment in children affected with AIS.

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