Genetic Steroid Disorders: Chapter 8. The Persistent Müllerian Duct Syndrome

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The persistent Müllerian duct syndrome (PMDS) is characterized by the persistence of Müllerian derivatives in otherwise normally virilized XY individuals. The condition is usually due to a mutation in either the anti-Müllerian hormone (AMH) or the AMH type II receptor (AMHR-II) genes and is transmitted as a recessive autosomal trait. Sixty-five families with AMH mutations and 59 with AMHR-II mutations have been reported to date. Clinical symptoms include cryptorchidism and/or inguinal hernia, and are identical for ligand and receptor mutations. However, the prepubertal serum level of AMH is nearly undetectable in AMH mutations, whereas it is close to normal in receptor mutations. Infertility is the main complication. Construction of molecular models for the AMH and AMHR-II has provided insight into how some mutations affect the biosynthesis and processing of these molecules, and how other mutations affect signal transduction.

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