Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency.
ORPHA:90791

Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency is a very rare form of congenital adrenal hyperplasia (CAH; see this term) encompassing salt-wasting and non-salt wasting forms with a wide variety of symptoms, including glucocorticoid deficiency and male undervirilization manifesting as a micropenis to severe perineoscrotal hypospadias.