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Review Article

Anti-Müllerian Hormone and Its Clinical Use in Pediatrics with Special Emphasis on Disorders of Sex Development

Marie Lindhardt Johansen,[1] Casper P. Hagen,[1] Trine Holm Johannsen,[1] Katharina M. Main,[1] Jean-Yves Picard,[2] Anne Jørgensen,[1] Ewa Rajpert-De Meyts,[1] and Anders Juul[1]

Using measurements of circulating anti-Müllerian hormone (AMH) in diagnosing and managing reproductive disorders in pediatric patients requires thorough knowledge on normative values according to age and gender. We provide age- and sex-specific reference ranges for the Immunotech assay and conversion factors for the DSL and Generation II assays. With this tool in hand, the pediatrician can use serum concentrations of AMH when determining the presence of testicular tissue in patients with bilaterally absent testes or more severe Disorders of Sex Development (DSD). Furthermore, AMH can be used as a marker of premature ovarian insufficiency (POI) in both Turner Syndrome patients and in girls with cancer after treatment with alkylating gonadotoxic agents. Lastly, its usefulness has been proposed in the diagnosis of polycystic ovarian syndrome (PCOS) and ovarian granulosa cell tumors and in the evaluation of patients with hypogonadotropic hypogonadism.

Review Article

Anti-Müllerian Hormone: A Valuable Addition to the Toolbox of the Pediatric Endocrinologist

Nathalie Josso,[1] Rodolfo A. Rey,[2,3] and Jean-Yves Picard[1]

Anti-Müllerian hormone (AMH), secreted by immature Sertoli cells, provokes the regression of male fetal Müllerian ducts. FSH stimulates AMH production; during puberty, AMH is downregulated by intratesticular testosterone and meiotic germ cells. In boys, AMH determination is useful in the clinical setting. Serum AMH, which is low in infants with congenital central hypogonadism, increases with FSH treatment. AMH is also low in patients with primary hypogonadism, for instance in Down syndrome, from early postnatal life and in Klinefelter syndrome from midpuberty. In boys with nonpalpable gonads, AMH determination, without the need for a stimulation test, is useful to distinguish between bilaterally abdominal gonads and anorchism. In patients with disorders of sex development (DSD), serum AMH determination helps as a first line test to orientate the etiologic diagnosis: low AMH is indicative of dysgenetic DSD whereas normal AMH is suggestive of androgen synthesis or action defects. Finally, in patients with persistent Müllerian duct syndrome (PMDS), undetectable serum AMH drives the genetic search to mutations in the AMH gene, whereas normal or high AMH is indicative of an end organ defect due to AMH receptor gene defects.