Early onset of primary hypogonadism revealed by serum anti-Müllerian hormone determination during infancy and childhood in trisomy 21

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Male patients with an extra sex chromosome or autosome are expected to present primary hypogonadism at puberty owing to meiotic germ-cell failure. Scarce information is available on trisomy 21, a frequent autosomal aneuploidy. Our objective was to assess whether trisomy 21 presents with pubertal-onset, germ-cell specific, primary hypogonadism in males, or whether the hypogonadism is established earlier and affects other testicular cell populations. We assessed the functional status of the pituitary-testicular axis, especially Sertoli cell function, in 117 boys with trisomy 21 (ages: 2months-20year). To compare with an adequate control population, we

established reference levels for serum anti-Müllerian hormone (AMH) in 421 normal males, from birth to adulthood, using a recently developed ultrasensitive assay. In trisomy 21, AMH was lower than normal, indicating Sertoli cell dysfunction, from early infancy, independently of the existence of cryptorchidism. The overall prevalence rate of