

Macroorchidism In Childhood and Adolescence: an Update

Vincenzo De Sanctis MD¹, Maria Marsella MD²,
Ashraf Soliman, MD³, PhD, FRCP

¹Pediatric and Adolescent Outpatient Clinic, Quisisana Hospital, 44100 Ferrara, Italy, ²Hospital of Sacred Heart of Jesus, Benevento, Italy, ³Departments of Pediatrics and Hematology, Hamad General Hospital, Doha, Qatar

Corresponding author: Vincenzo De Sanctis MD, Pediatric and Adolescent Outpatient Clinic, Quisisana Hospital, 44100 Ferrara, Italy; Tel:39 0532 770243 ,Email: vdesantis@libero.it

Abstract

In the last 30 years, scientific literature has been enriched with studies which demonstrate the importance of evaluating testis volume to recognize certain genetic and endocrine diseases. Macroorchidism is defined as an increase of testicular volume at least twice the norm for age. In clinical practice, when macroorchidism is suspected, the testicular volume may be evaluated with Prader' orchidometer and/or US, calculated by this formula: $L \times W \times H \times 0.71$ and the resulting value should be compared with a table of percentiles for each age. Macroorchidism is a relatively uncommon sign; however, when present, it almost always has clinical relevance. Five groups of conditions are reviewed: genetic, endocrine, idiopathic and secondary to neoplasm or testicular torsions. An X-linked mental retardation syndrome must be suspected in all patients with macroorchidism, phenotypic abnormalities, and mental retardation. Furthermore, it is important to identify other males affected in the same family to confirm the X-linked transmission. In these cases, the patient must be referred to specialized cytogenetic centres for determination of fragile sites. Other possible etiologies of macroorchidism are long-standing primary hypothyroidism, adrenal remnants in congenital adrenal hyperplasia, follicle stimulating hormone (FSH)-secreting pituitary macroadenomas, local tumors, lymphomas, and aromatase deficiency. Early diagnosis is

important in order to identify and reduce the incidence of X-linked mental retardation in affected families and to begin treatment in endocrinologic, tumoral and surgical disorders.

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