Child With Suspected Delayed Puberty

Suggestive history and physical findings	Initial laboratory and/or radiologic work-up can include:	When to refer	Items useful for consultation	Additional information
Symptoms/Signs: Boys: No testicular enlargement (≥4ml or 2.5 cm in long diameter) by age 14 years Girls: No breast development by age 13 years Boys and girls: • may also have delay in adrenarche - Pubic hair, axillary hair, body odor and acne • also look for CNS symptoms, chronic disease symptoms (IBD, CF, SSD, asthma) **Family history:** constitutional delay Differential Diagnosis	**Radiological tests:** • Bone age **Blood tests:*** • LH • FSH • testosterone (males) • estradiol (females) • TSH • Free T4 • prolactin *Pubertal laboratory tests should be obtained in the early AM using sensitive pediatric assays only Other tests to consider after consultation with Pediatric Endocrinologist: • Pelvic ultrasound • Chromosome analysis	**Urgent:** if CNS symptoms present; Turner syndrome diagnosed **Routine:** All others Find a Pediatric Endocrinologist	Previous growth data/growth charts Pertinent medical records Recent laboratory and radiologic studies (including actual copy of bone age)	Delayed Puberty in Boys: A Guide for Families Delayed Puberty in Girls: A Guide for Families References

Differential diagnosis for delayed puberty:

- Constitutional delay (53%)
- Functional hypogonadotropic hypogonadism (19%)
    - Chronic illness, malnutrition, overtraining, hypothyroidism, growth hormone deficiency
- Hypogonadotropic hypogonadism (HH) (12%)
    - Intracranial disorders including tumors and congenital abnormalities
    - Isolated gene defects such as DAX1, Kallman Syndrome, GnRH/receptor
    - Hypopituitarism
    - Part of a syndrome: Prader-Willi, Bardet-Biedl, Laurence-Moon
    - Permanent damage to hypothalamus/pituitary due to secondary disease i.e. iron overload
- Hypergonadotropic hypogonadism (13%)
    - Abnormal sex chromosomes: Turner syndrome, Klinefelter Syndrome
    - Damage to gonads: trauma, torsion, chemotherapy, radiation, galactosemia, iron overload, cystic fibrosis, mumps orchitis, anorchia, cryptorchidism
    - Disorders of sex differentiation: androgen insensitivity, gonadal dysgenesis
- Unclassified (3%).

Suggested Reference and Additional Reading:

- Wei C, Crowne EC. Recent advances in the understanding and management of delayed puberty. Arch Dis Child 2016 May;101(5):481-8.

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