

Abnormal pubertal development

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Rationale

Puberty is the physiologic and psychosocial transition from childhood to adolescence. Questions about typical and atypical pubertal development are a common reason for presentation to primary care clinics. Abnormalities in pubertal development can be indicators of severe underlying disorders and can be a cause of significant anxiety for patients and families. They require careful investigation and follow-up.

Causal Conditions

(list not exhaustive)

- Delayed puberty
 - a. Variant of normal (idiopathic constitutional delay of puberty)
 - b. Primary gonadal disorders
 - Congenital
 - Chromosomal (e.g., Turner syndrome, Klinefelter syndrome)
 - Congenital malformations
 - Acquired gonadal disorders (e.g., gonadal infection, trauma, neoplasm)
 - c. Secondary gonadal disorders
 - Functional (e.g., chronic illness, malnutrition)
 - Hypothalamic dysfunction (e.g., hyperprolactinemia, exogenous steroids)
 - Pituitary dysfunction (e.g., central nervous system [CNS] tumour)
- Precocious puberty

- a. Central precocious puberty (gonadotropin-dependent)
 - Idiopathic
 - CNS (e.g., neoplasms, hydrocephalus)
 - b. Peripheral precocious puberty (gonadotropin-independent)
 - Autonomous gonadal function (e.g., ovarian cysts, Leydig cell tumours of ovaries or testes)
 - Adrenal pathology (e.g., tumours, congenital adrenal hyperplasia)
 - Exogenous sex hormone exposure
- Variations of early pubertal development (e.g., premature thelarche, premature adrenarche)

Key Objectives

Given a patient in whom there are concerns about pubertal development, the candidate will identify the cause, severity, and complications, and will initiate an appropriate management plan. Particular attention should be paid to distinguishing normal variation of pubertal development from symptoms of serious underlying disorders, and to supportive counselling regarding the psychosocial aspects of puberty.

Enabling Objectives

Given a patient in whom there are concerns about pubertal development, the candidate will

- list and interpret relevant clinical findings, including those derived from
 - a. an appropriate history, with a focus on growth and development, nutrition, and symptoms of underlying systemic disease; and
 - b. an appropriate physical examination, with particular attention to Tanner staging of pubertal development and to signs of underlying disorders (e.g., CNS tumours, eating disorders);
- list and interpret relevant investigations, including differentiation of normal variants from serious or urgent underlying conditions (e.g., CNS or pelvic imaging if neoplasm is suspected); and
- construct an effective initial management plan, including
 - a. reassurance in case of normal variants of pubertal development;

- b. referral for appropriate specialized care (e.g., pediatrics, endocrinology, genetics, neurology) in case of abnormal pubertal development; and
- c. supportive counselling to the patient and their family regarding the psychosocial implications of abnormal pubertal development.