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Disorders of puberty and polycystic ovary syndrome (PCOS)

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Polycystic ovary syndrome (PCOS) is a heterogeneous familial disorder characterized by chronic anovulation and hyperandrogenism (clinical and/or biochemical). This multi-system, polygenic, multi-factorial disorder is associated with an increased risk for metabolic abnormalities such as type 2 diabetes mellitus. Signs and symptoms of PCOS often emerge during the peri-pubertal years with premature pubarche being the earliest manifestation for some girls. Central obesity, Insulin resistance and hyperinsulinemia are important pathophysiological features that are common to both premature pubarche and PCOS. Increasing evidence suggests that PCOS arises as a complex trait with contributions from both heritable and nonheritable factors. Polycystic ovaries appear to be transmitted as a dominant trait, usually asymptomatic but often accompanied by a subclinical PCOS type of ovarian dysfunction. Risk factors such as premature pubarche, obesity, ethnicity, and family history may be helpful. The evaluation of the adolescent with suspected hyperandrogenism or PCOS must be individualized depending on the symptoms and examination findings. Treatment must be individualized, too, and often requires a multidisciplinary approach.

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