

DELAYED PUBERTY

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Since puberty is a long ongoing developmental process with significant individual and population differences in timing, the definition of delayed puberty for a given individual is based on arbitrary criteria given by epidemiological data.

Delayed or late puberty is discussed when there is no breast development at an age that is 2 to 2.5 SD later than the mean age at which this event occurs i.e. 13 years.

The differential diagnosis between delayed puberty and absence of puberty i.e. hypothalamo-pituitary defects or ovarian failure is still puzzling: there is no gold-standard test with a sufficiently high degree of sensitivity/specificity that could allow one to differentiate:

- variability of pubertal development

- hypogonadotropic hypogonadism (genetically or functional).

Late puberty can affect psychosocial wellbeing of adolescent girls: beside anxiety and depression symptoms, decreased self-esteem, concerns regarding development of sexual identity and fertility are frequently reported. Psychological support should be systematically discussed.

First line evaluation includes:

- family history
- physical examination (Tanner stage, growth rate)
- hormonal analyses (plasma level of FSH, LH, PRL, IGF-1)
- imaging (bone age, pelvic US).
- Second line investigation associates:
- GnRH stimulating test
- olfactory function test
- brain MRI

- genetic and molecular genetic analyses.

Diagnosis of a girl with delayed/absence of puberty includes:

1. Constitutional delayed puberty (and growth) (CDPG): about 25%.

No test can reliably distinguish CDPG from isolated hypogonadotropic hypogonadism (IHH). IHH is diagnosed if puberty is not begun by the age of 18 years.

2. Hypogonadotropic hypogonadism: about 45%.

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It can be:

- acquired: post-tumoral, post-radiotherapy

- functional: stress, anorexia, sport

- congenital: isolated GnRH deficiency.

3. Hypergonadotropic hypogonadism: 30%.

It can be:

- acquired: post-chemotherapy, ...

- congenital: XO (Turner), XX (premature ovarian insufficiency), or XY (DSD).

Treatment should be discussed, according to the cause of delayed/absence of puberty, in agreement with the adolescent personal choice.