Delayed Puberty

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Aims/Objectives

- By the end of this session, you should be able to…
- Describe what delayed puberty is
- Explain the physiology of normal onset of puberty and its clinical picture
- Recognize the differences between the two forms of organic delayed puberty
- Discuss the investigations that are needed to aid diagnosis of delayed puberty

Describe What Delayed Puberty Is

Definition

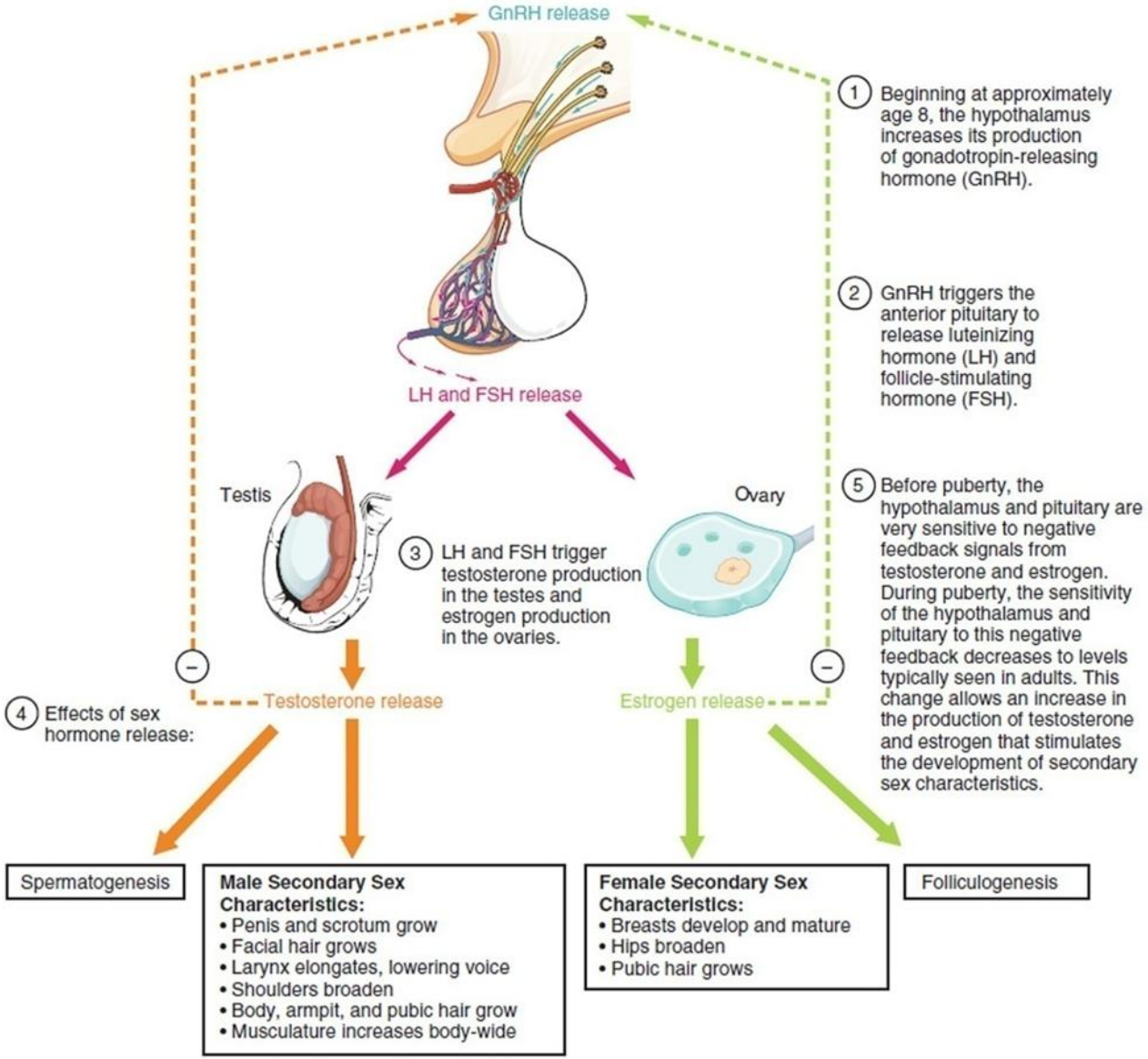
- Defined as the lack of any pubertal signs by the age of:
- 13 years in girls
- 14 years in boys

- Boys = Absence of testicular enlargement (<4mls) by 14 years
- Girls = Absence of breast budding by 13 years

- More common in boys

Explain The Physiology of Normal Onset of Puberty And Its Clinical Picture

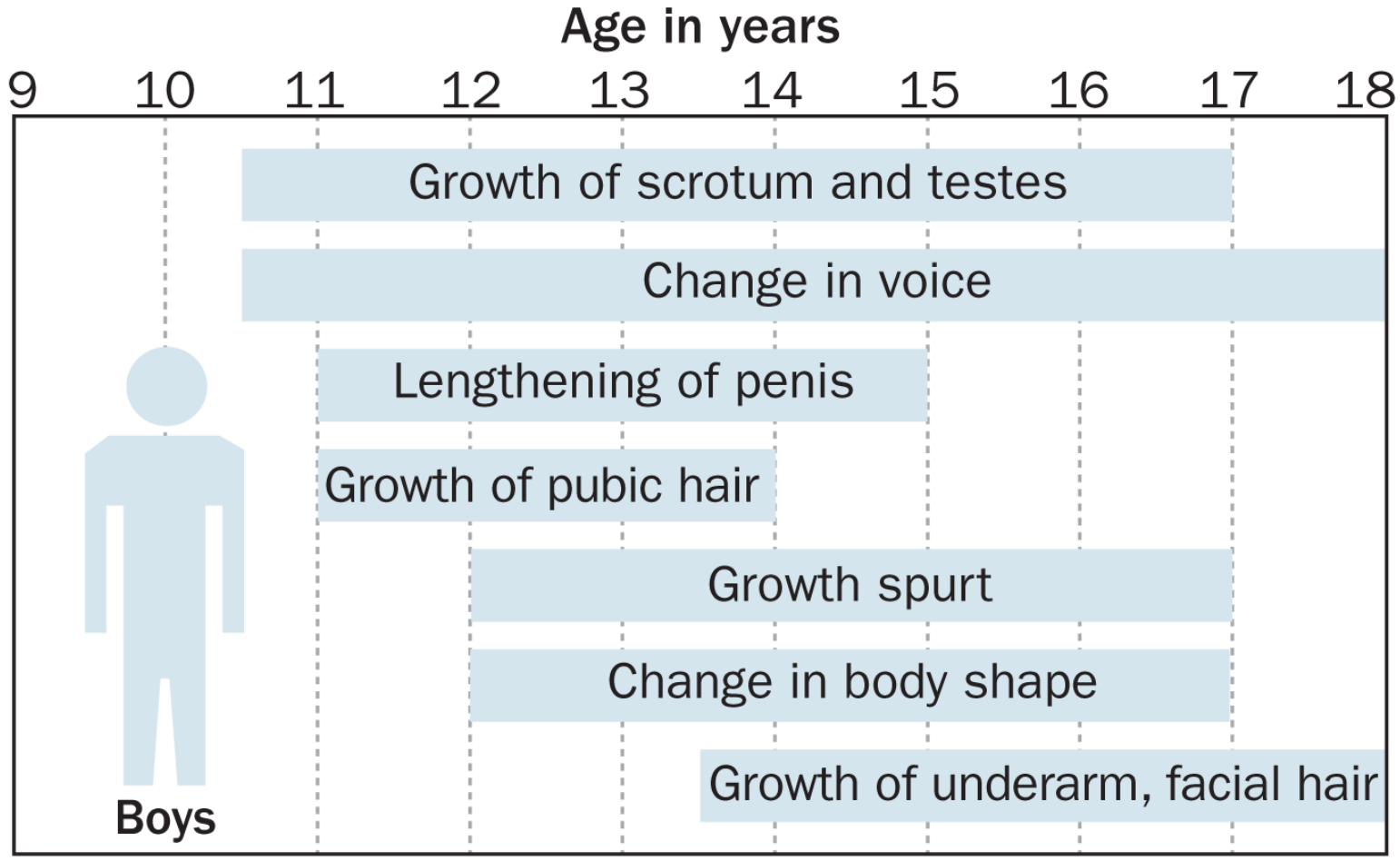
Physiology – Normal Puberty



Physiology - Normal pubertal development

- Boys –
- First sign of puberty is increase in the size of the testes
- Followed by penile and scrotal changes
- Testicular size is documented as a measurement of testicular volume using the Prader orchidometer.
- Volume of 4 mL defines the onset of puberty
- Axillary hair/voice/increase in growth velocity only occur in mid to late puberty
- Growth spurt at Tanner Stage 3-4
- Facial hair does not appear until late puberty

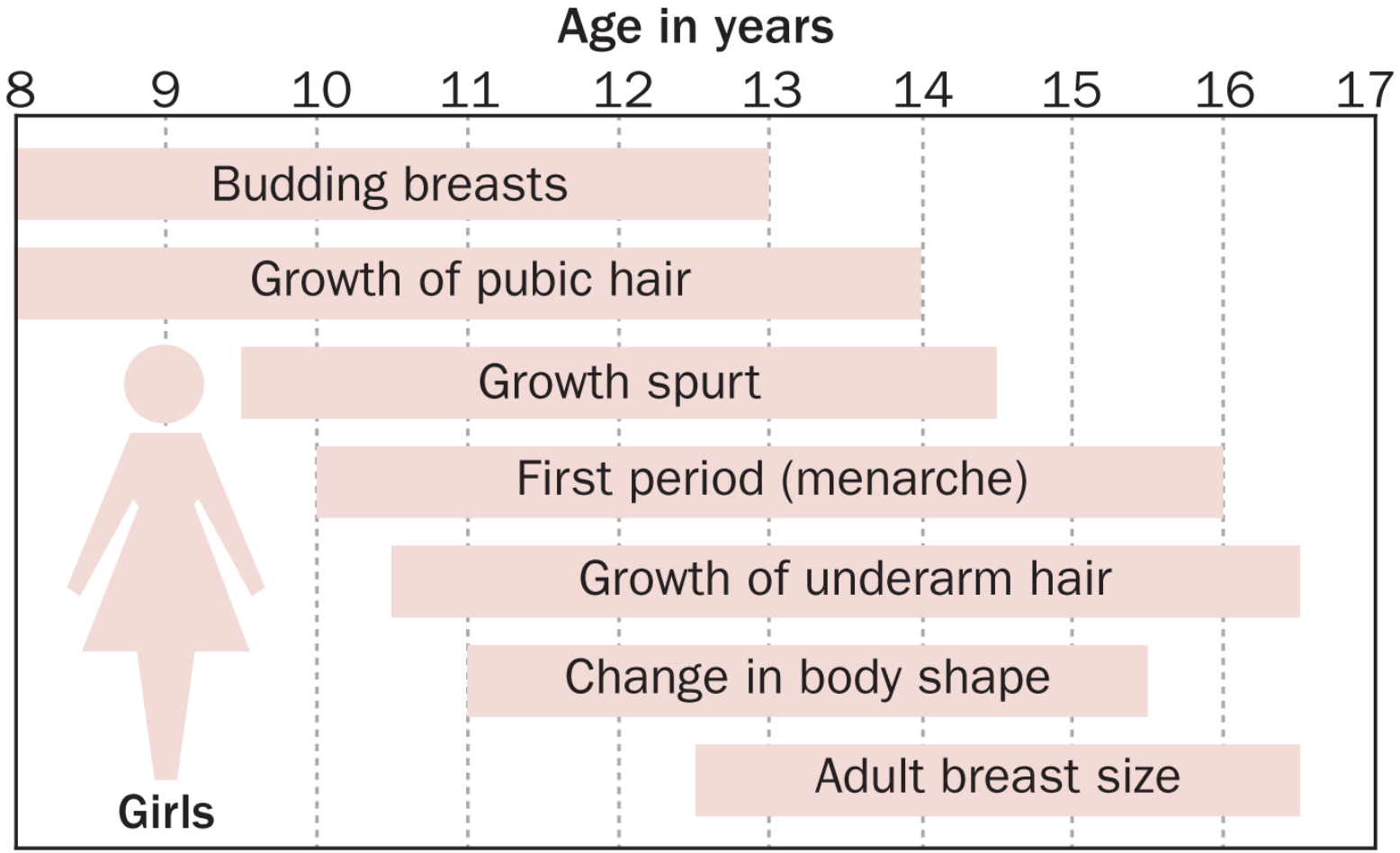
Physiology - Normal pubertal development



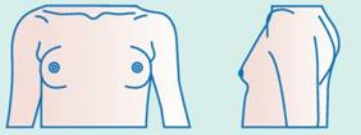
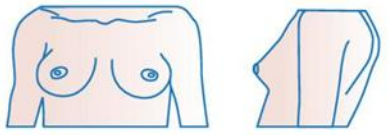
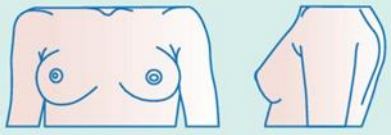
Physiology - Normal pubertal development

- Girls -
- First sign of puberty is breast development
- Pubic and axillary hair, acne, and body odour develop as a result of androgens secreted from the adrenal gland
- Peak growth spurt occurs in Tanner stage 2-3
- Menarche occurs at Tanner stage 4 breast development

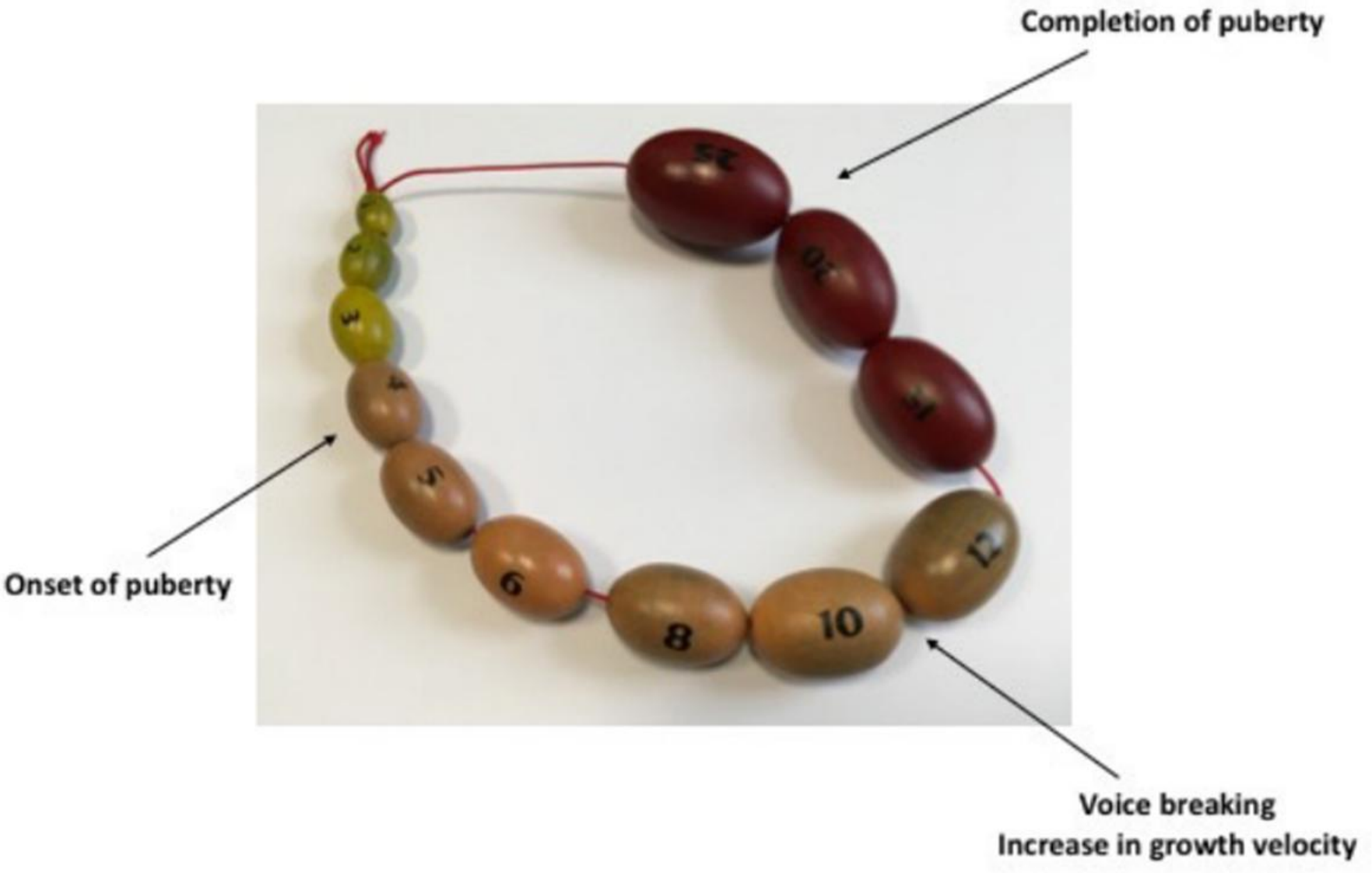
Physiology - Normal pubertal development



Examination

Tanner stage	Male genital appearance	Male genital description	Female pubic hair appearance	Pubic hair description	Breast appearance	Breast description
1		Testicular volume <3ml		No pubic hair		Elevation of papilla only
2		Testicular volume <3ml, change in texture to scrotal skin		Sparse growth chiefly along the labia/base of penis		Breast bud stage
3		Increase in size of penis with further testicular enlargement		Darker, coarser, more curled hair		Enlargement of breast and areola
4		Further enlargment of penis and testicles with development of glans penis		Adult type hair over a smaller area		Projection of the areola and papilla
5		Adult size and shape		Spread to the medial surface of the thighs		Recession of the areola to the contour of the breast, projection of papilla only

Examination



Recognize The Differences Between The Two Forms Of Organic Delayed Puberty

Causes

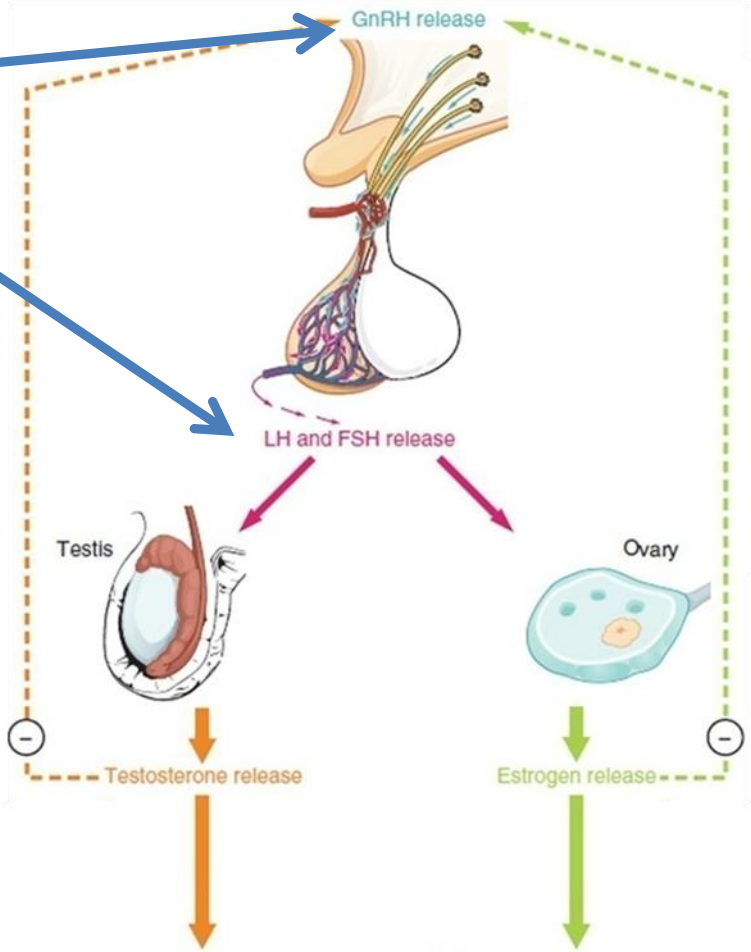
- Functional:
  - Constitutional delay
  - Underlying chronic disease e.g. CF, Coeliac, IBD, Poorly controlled T1DM
  - Malnutrition e.g. Anorexia
  - Excessive exercise
- Due to physiological delay in HPG axis activation without associated pathology
- Often familial, most common in boys
- Associated with short stature that is appropriate for the skeletal age

Causes

- Organic:
- Hypogonadotrophic Hypogonadism -
  - Due to lack of serum gonadotrophin production
- Hypergonadotrophic Hypogonadism -
  - Due to gonadal insufficiency with elevated gonadotrophins

Hypogonadotrophic Hypogonadism

Problem here



Lack of sex steroid production

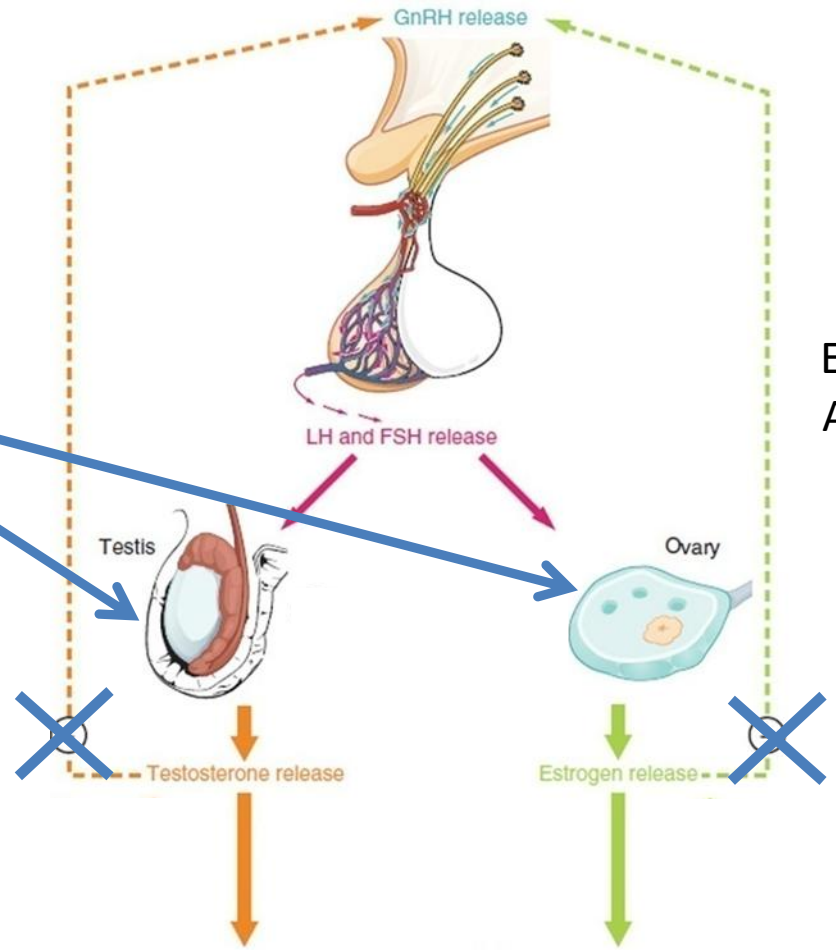
Either will cause low LH+FSH

This will not distinguish between Hypogonadotrophic Hypogonadism and functional causes

Hy**po**gonadotrophic Hypogonadism

- Hypogonadism due to problems with either the hypothalamus or pituitary gland - affecting the (HPG axis)
- Hypothalamic disorders = result from a deficiency in the release of gonadotropic releasing hormone (GnRH)
- Pituitary gland disorders = due to a deficiency in the release of gonadotropins from the anterior pituitary

Hypergonadotrophic Hypogonadism



Problem here

Elevated gonadotropin levels
An attempt of compensation by the body

= Raised LH+FSH

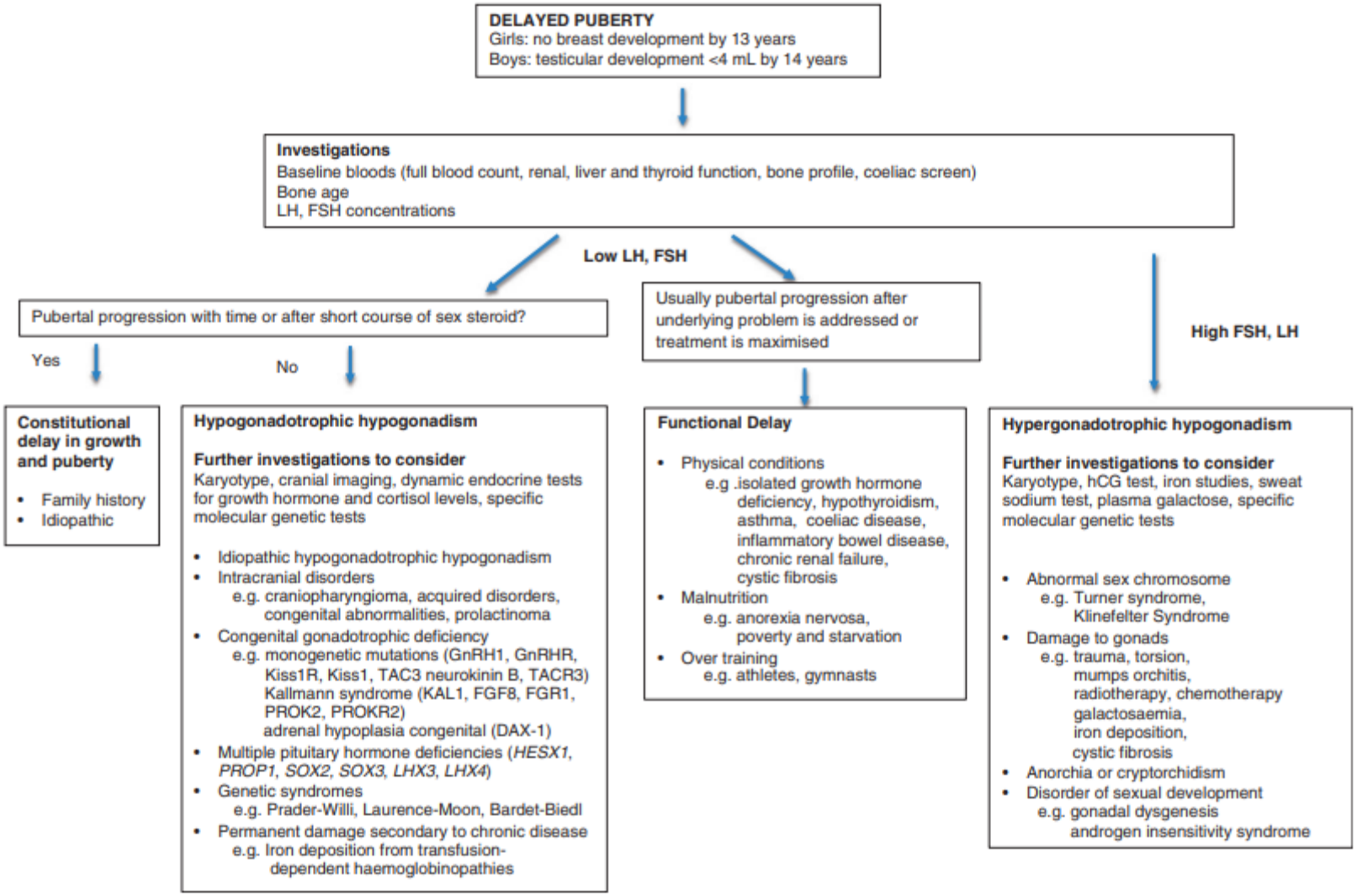
Lack of sex steroid production

Hypergonadotrophic Hypogonadism

- Hypogonadism due to an impaired response of the gonads to the gonadotropins (FSH and LH)
- This results in:
  - A lack of sex steroid production
  - Elevated gonadotropin levels (as an attempt of compensation by the body)
- Caused by:
- Congenital –
  - Chromosomal abnormalities (resulting in gonadal dysgenesis) - Turner's syndrome, Klinefelter's syndrome
  - Gonadotropin resistance - Leydig cell hypoplasia (or insensitivity to LH) in males, FSH insensitivity in females
- Acquired - (due to damage to/dysfunction of the gonads)
  - Torsion, orchitis, premature ovarian failure, trauma, surgery, STIs, drugs (e.g., antiandrogens, opioids, alcohol)

Discuss The Investigations That Are Needed To Aid Diagnosis Of Delayed Puberty

Investigations



Treatment

- Boys:
- Testosterone monthly IM injection – dose of Testosterone increases every 6 months
- 50mg (i.e., 0.2ml of the 250mg/ml) – Monthly IM injection – increase by 25mg every 6 months up to highest of 150mg

- Girls:
- Oestrogen patches – size of patch increases every 6 months
- ¼ patch 3 days a week initially

Any Questions?

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References

- Prentice 2021. Fifteen-minute consultation: Clinical pubertal assessment. Arch Dis Child Educ Pract Ed 2022;107:253–256.
- Wei 2017. The investigations of children and adolescents with abnormalities of pubertal timing. Annals of Clinical Biochemistry 2017. Vol 54 (1):20-32.