

# ABNORMAL PUBERTY

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# Objectives

By the end of the lecture students will be able to:

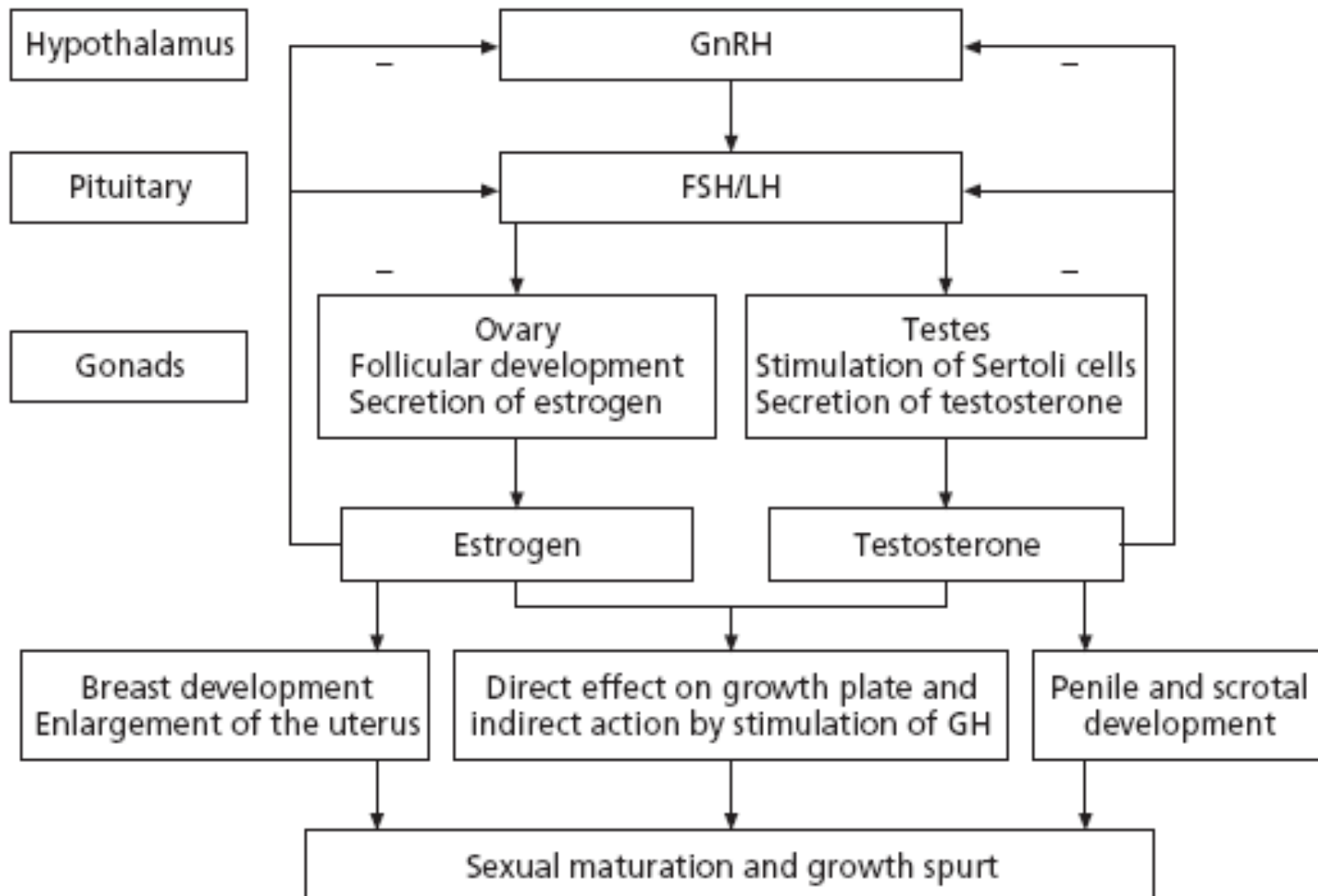
- definition of puberty
- Hormonal changes during normal puberty
- Normal pubertal stages in both sexes
- definition & causes of precocious puberty
- Definition & causes of delayed puberty

# Definition of puberty

- The stage between the onset of secondary sexual characteristics & completion of physical maturity
- The period in which reproductive capability is attained, manifested by spermatogenesis in males & ovulation in females

# Endocrine changes in puberty

PROBLEMS OF PUBERTY AND ADULESC

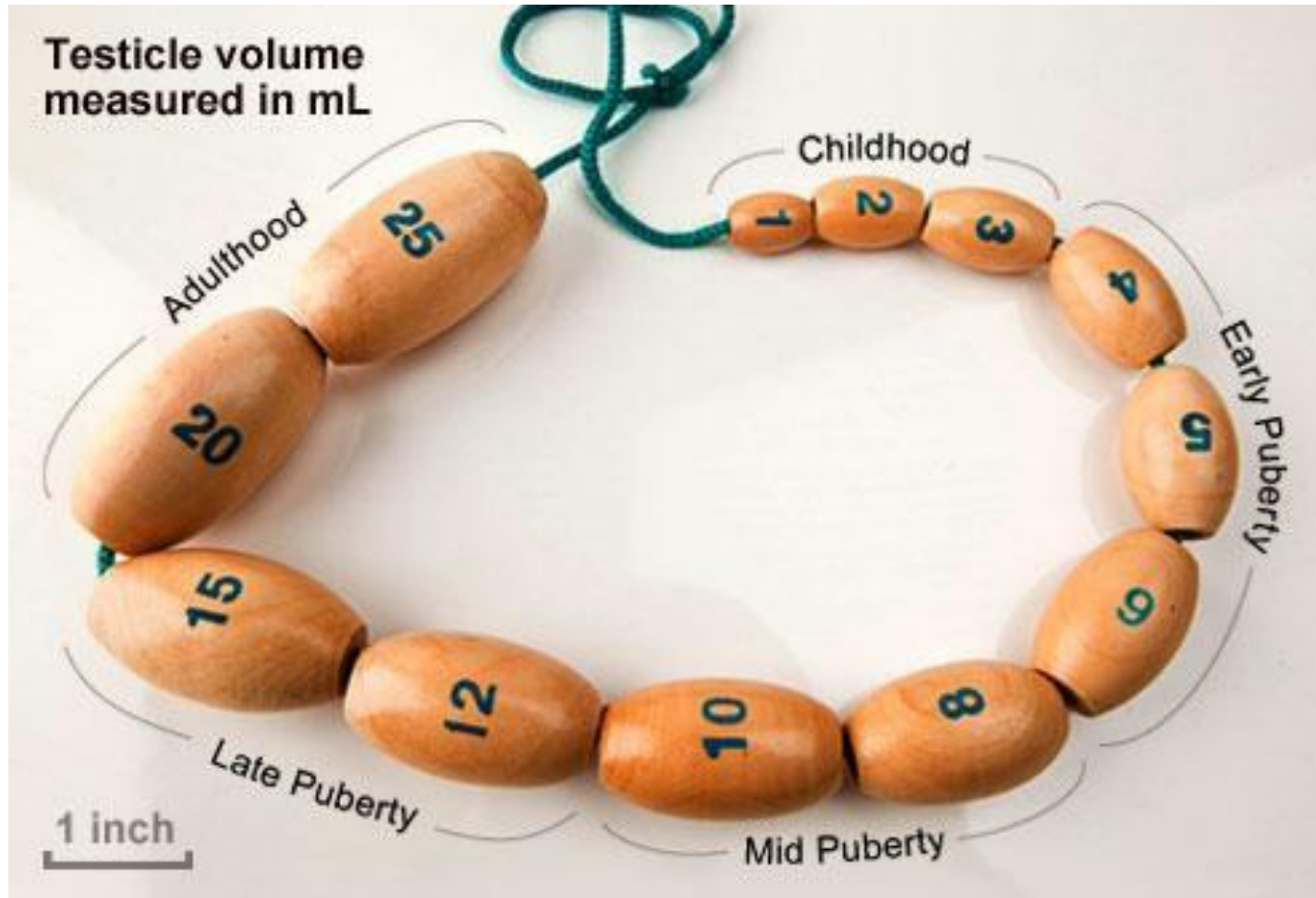


# Manifestations of puberty

## Puberty in boys

- First sign is testicular enlargement followed by pubic hair development and genital enlargement
- Puberty starts when the testes are 4 ml in size (measured by Orchidometer)
- Adult testes are 20-25 ml in size

# Orchidometer



# Somatic changes in boys

- Growth spurt is later than girls by 2 years
- Increased muscle mass
- Decreased adipose tissue
- Skeletal changes ( $\uparrow$ BMD)
- 60% have transient gynaecomastia
- Spermatogenesis average by 15 year

# Pubertal stages in Boys

## Genital development

- *Stage 1:* Preadolescent
  - The testes, scrotum and penis are of about the same size and proportions as in early childhood
- *Stage 2:* Enlargement of the scrotum and testes
  - The skin of the scrotum reddens and changes in texture. Little or no enlargement of the penis
- *Stage 3:* Lengthening of the penis
- *Stage 4:* Increase in breadth of the penis and development of the glans.
  - The testes and scrotum are larger; the scrotum darkens.
- *Stage 5:* Adult

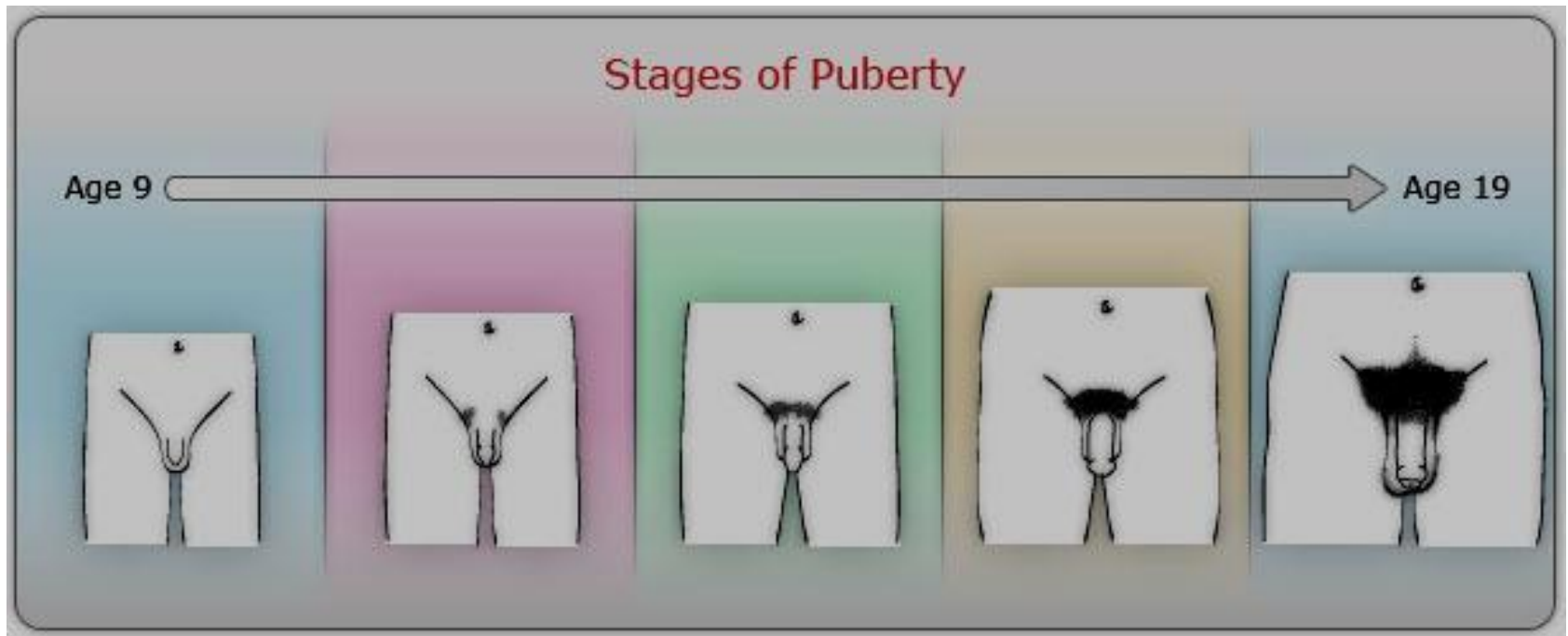


# Pubertal stages in Boys

## Pubic hair

- *Stage 1:* Preadolescent. No pubic hair.
- *Stage 2:* Sparse growth of slightly pigmented downy hair at the base of the penis.
- *Stage 3:* Hair darker, coarser and more curled, spreading sparsely over the junction of the pubes.
- *Stage 4:* Hair adult in type, but covering a considerably smaller area than in the adult. No spread to the medial surface of the thighs.
- *Stage 5:* Adult quantity and type and spread to the medial surface of the thighs.
  - Spread up linea alba occurs late, in about 80% of men, after adolescence is complete, and is rated Stage 6.

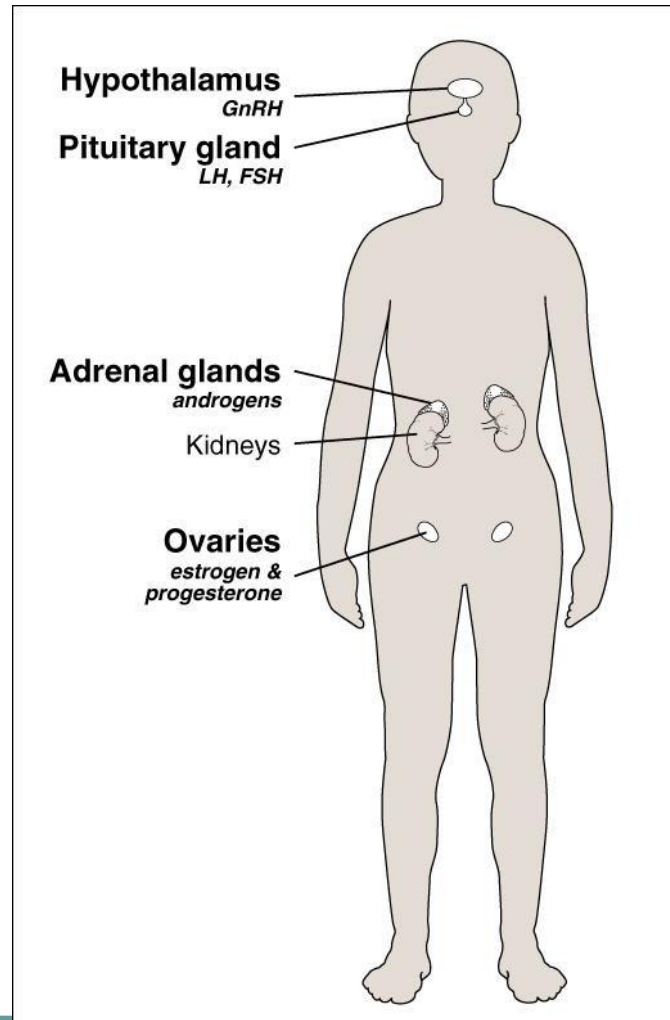
# Pubertal changes in boys



# Puberty in girls

- First sign is breast enlargement followed by pubic and axillary hair and lastly menstruation which is called menarche
- Pelvic U/S changes of puberty
  - ↑ovarian volume and follicular size
  - ↑uterus to cervix ratio
  - ↑endometrial echo

# Normal puberty in girls



# Skeletal changes in girls

- Widening of pelvis and carrying angle
- Major increase in BMD
- Increased adipose tissue with typical female distribution
- 95% of growth happened < menarche
- Menarche usually by age 11-14 year
- Increased in muscle bulk but not to same extent as males

# Pubertal stages in Girls

## Breast development

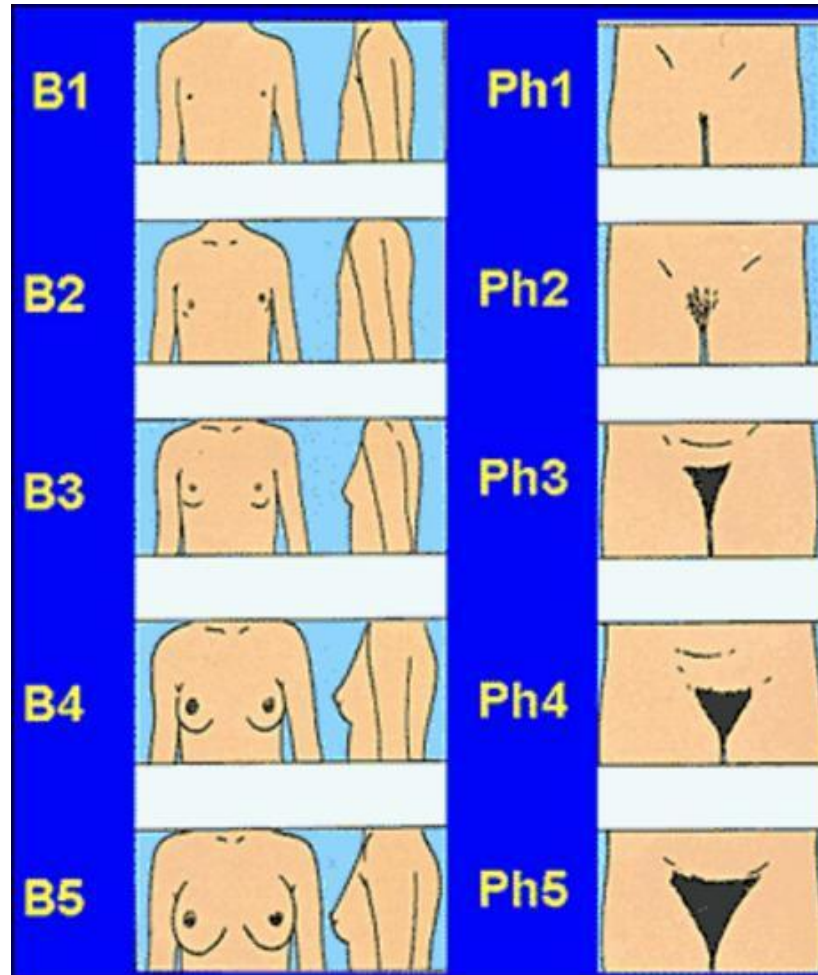
- *Stage 1:* Preadolescent.
- *Stage 2:* Breast bud stage. Elevation of the breast and papilla as a small mound.
  - Enlargement of the areola diameter.
- *Stage 3:* Further enlargement & elevation of the breast and areola, with no separation of their contours.
- *Stage 4:* Projection of the areola & papilla above the level of the breast.
- *Stage 5:* Mature stage, projection of the papilla alone due to recession of the areola.

# Pubertal stages in Girls

## Pubic hair

- *Stage 1:* Preadolescent. No pubic hair.
- *Stage 2:* Sparse growth of slightly pigmented downy hair along the labia.
- *Stage 3:* Hair darker, coarser and more curled, spreading sparsely over the junction of the pubes.
- *Stage 4:* Hair adult in type, but covering a considerably smaller area than in the adult. No spread to the medial surface of the thighs.
- *Stage 5:* Adult quantity and type with distribution of a horizontal pattern and spread to the medial surface of the thighs.
  - In about 10% of women, after adolescence is complete pubic hair spreads up the linea alba and is rated Stage 6.

# Pubertal changes in girls





# Precocious Puberty

## Definition

- In girls, defined as onset of puberty “breast enlargement” before age of 8 years
- In boys, defined as onset of puberty testicular enlargement before age of 9 years

# Types

- Central, True, GnRH dependent
  - 89-98% of cases (major type)
- Peripheral, Pseudo, GnRH Independent
  - 10 – 15 % of cases (not major type)
- Isolated Forms
  - Thelarche
  - Adrenarche / Pubarche

# Central Precocious Puberty

- Result from premature activation of Hypothalamus-Pituitary-Gonadal axis
- The pulsatile GnRH secretion leads to pulsatile secretions of LH and FSH with subsequent release of sex steroids
- Similar to normal mechanism but happened earlier than expected age

# Central, True, GnRH dependent

## Etiology

- Idiopathic
  - most girls ( 90 %)
- Secondary
  - most boys ( 70-80%)

# Etiology of Central type

## CNS disorders

- Hypothalamic Hamartoma
- Glioma (NF-1)
- Astrocytoma
- Craniopharyngioma
- Ependymoma, germinoma,
- CNS radiation therapy
- Post trauma (surgery)

# Etiology of Central type

- Inflammation (Brain abscesses)
- Neurological & mental retardation
- Hydrocephalus
- Prolonged primary hypothyroidism

# Etiology of peripheral type

- Gonadal: McCune-Albright, tumour, cyst
- Adrenal: Virilizing CAH, tumours
- Ectopic: hCG secreting tumours
  - Germinoma, Hepatoblastoma
- Exogenous source of hormone
- Familial male dependent (Testotoxicosis)

# Exogenous source of estrogens





# Delayed Puberty

## Definition:

- girls:
  - lack of breast development by age 13
  - more than five years between breast growth and menstrual period
  - lack of pubic hair by age 14
  - failure to menstruate by age 16
- boys:
  - lack of testicular enlargement by age 14
  - lack of pubic hair by age 15
  - more than five years to complete genital enlargement

# Types

- Two major types
  - Hypogonadotrophic hypogonadism
    - Hypothalamic -Pituitary defects
  - Hypergonadotrophic hypogonadism
    - Gonadal failure

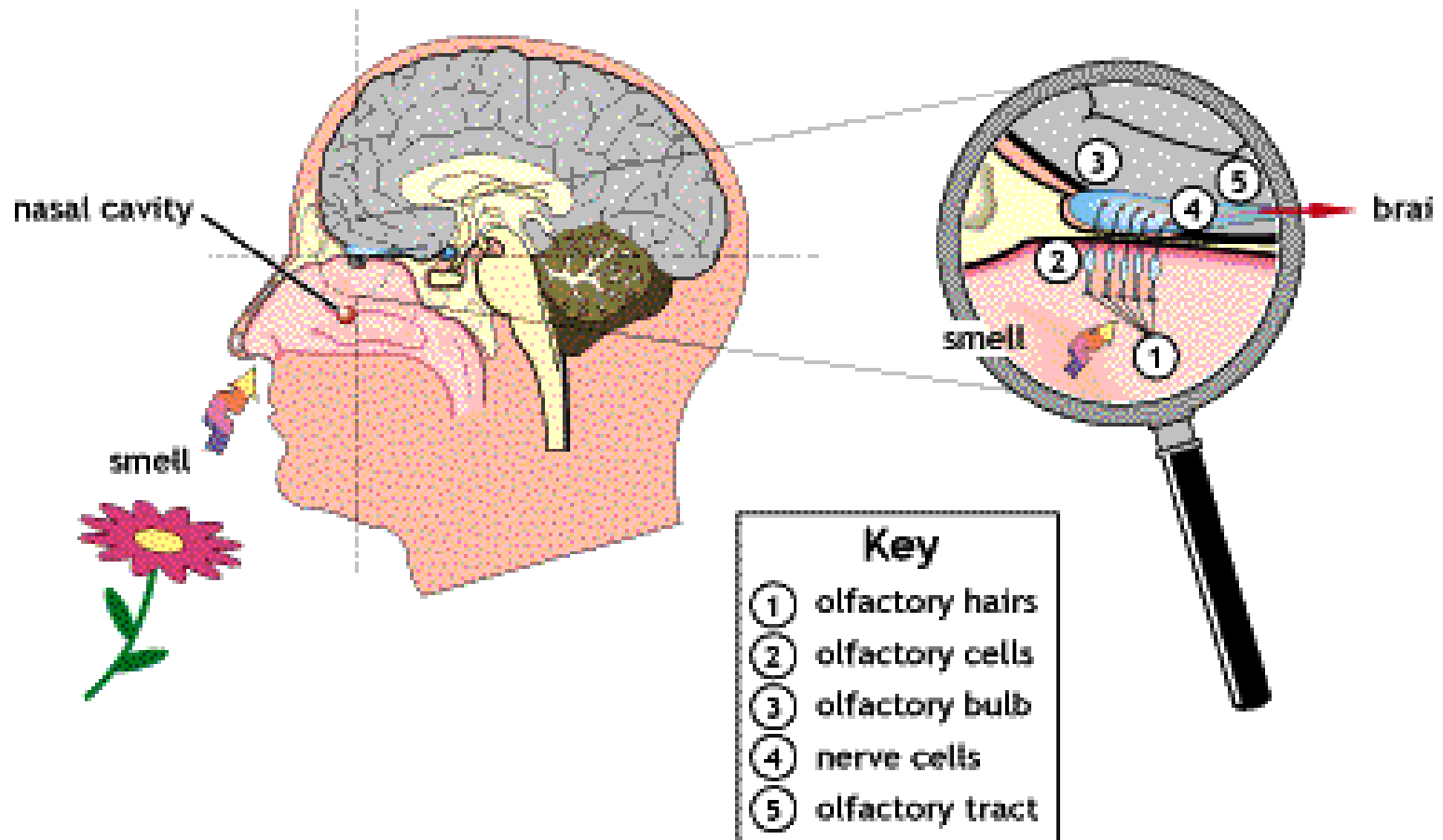
# Causes

- Constitutional delay
  - Commonest cause (~90% of cases)
  - Affecting both growth and puberty
  - Much more common (~10 fold) in boys than girls and may be familial
  - All investigations are normal

# Hypogonadotrophic hypogonadism

- Rare (~10%)
- Hypothalamic deficiency
  - GnRH deficiency - may be isolated or associated with other features e.g. anosmia (Kallman's syndrome), cognitive impairment and dysmorphic features (Prader-Willi syndrome)
- Pituitary deficiency
  - Gonadotrophin deficiency - may be isolated (LH deficiency) or more commonly associated with any form of hypopituitarism

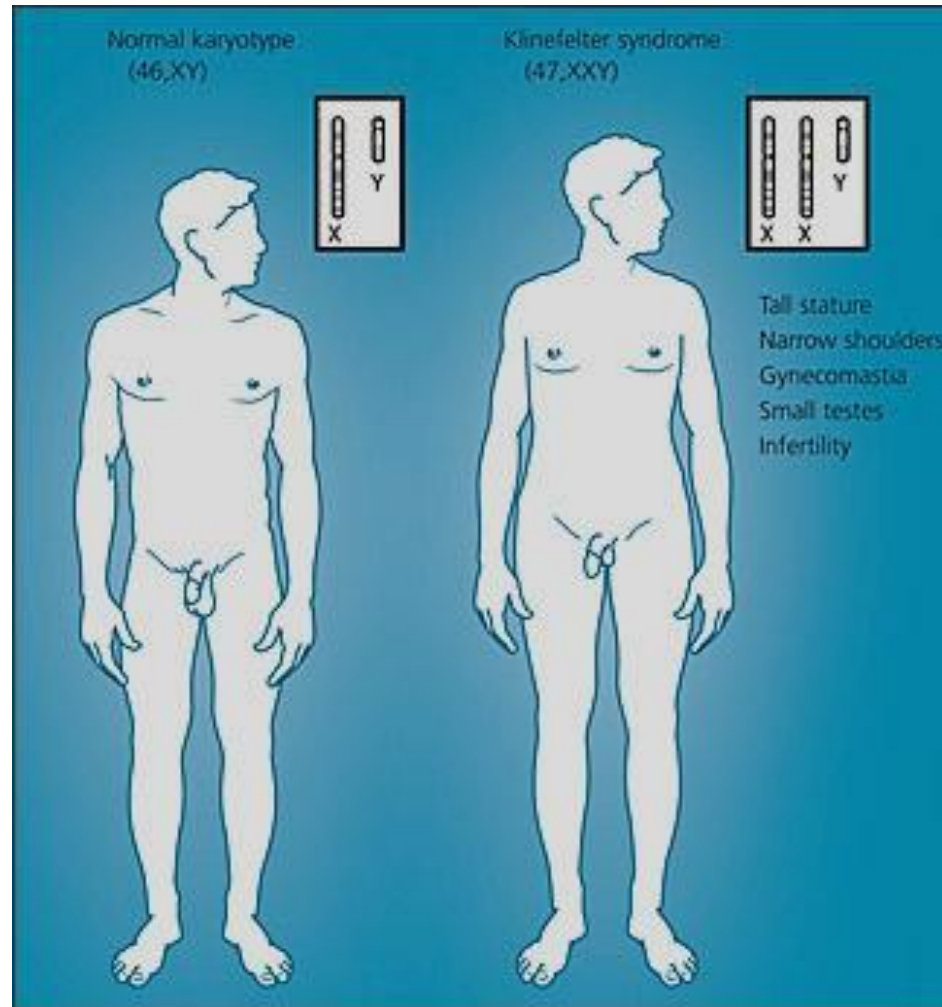
# Kallmann syndrome



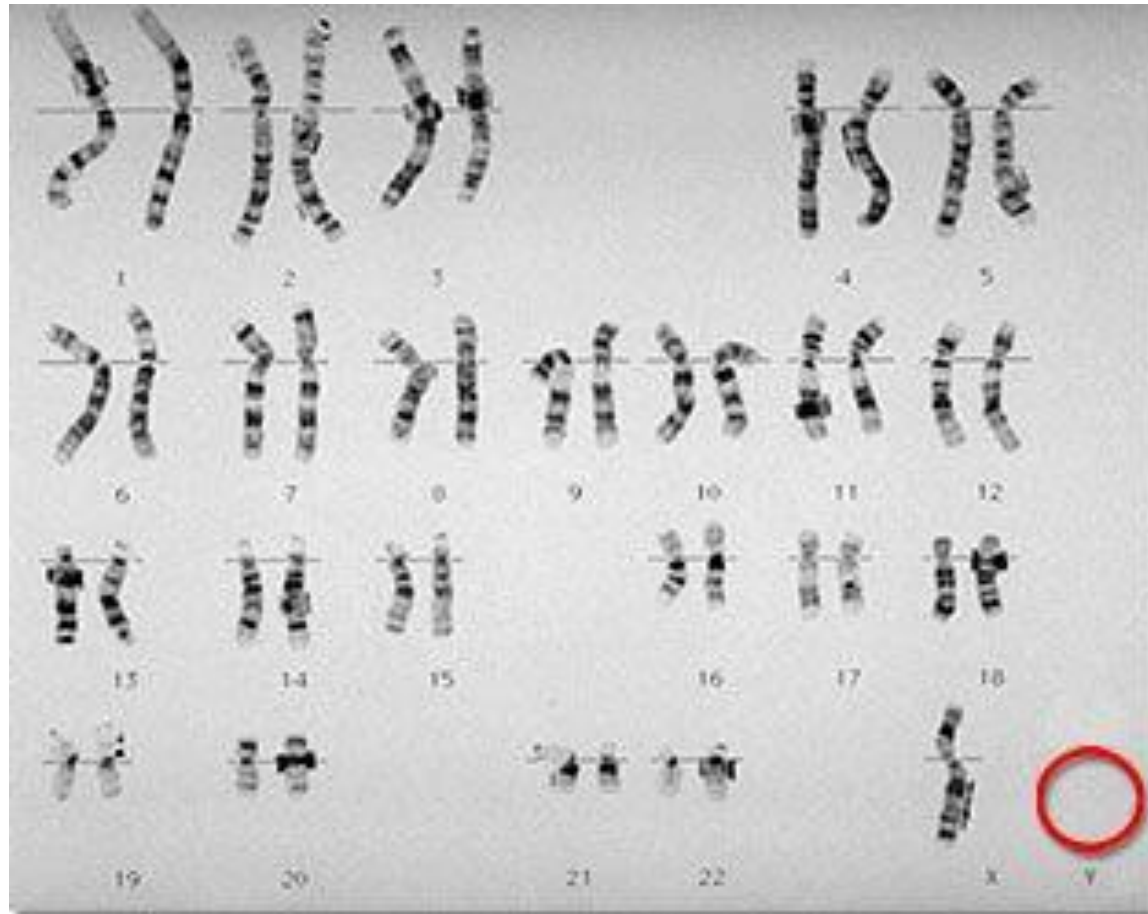
# Hypergonadotrophic hypogonadism

- Sex chromosome abnormalities
  - Klinefelter's syndrome in boys (47XXY)
  - Turner's syndrome in girls (45XO)
- Gonadal dysgenesis with normal Karyotype
- Gonadal damage
  - viral (e.g. mumps Orchitis)
  - iatrogenic (surgical, chemotherapy or radiotherapy)
  - autoimmune destruction (often associated with other autoimmune disease).
  - Gametes generally more sensitive to damage than steroid secreting cells

# Klinefelter syndrome



# Turner syndrome



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# Chronic Systemic Diseases

- Delay in pubertal development is very common in the presence of any serious illness e.g. chronic renal failure, bowel or liver diseases
- Progress depends on the course of the underlying disease
- Endocrine causes of delay puberty include hypothyroidism, GH deficiency and excess glucocorticoid

# Learning Points (1)

- Normal puberty starts in girls between 9 and 11 years, while in boys between 11 - 13 years
- Male puberty begins about 2 years later than girls
- Precocious puberty (before 8 years in girls, and before 9 years in boys)
- Precocious puberty is more common in girls than boys and is usually idiopathic

