Pediatric Endocrinology Review MCQs
(Part-1)

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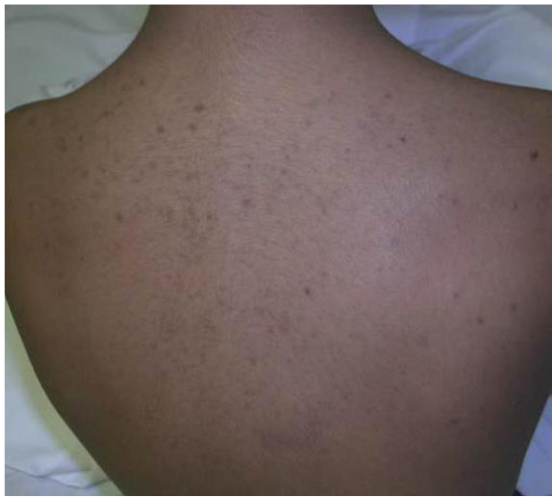
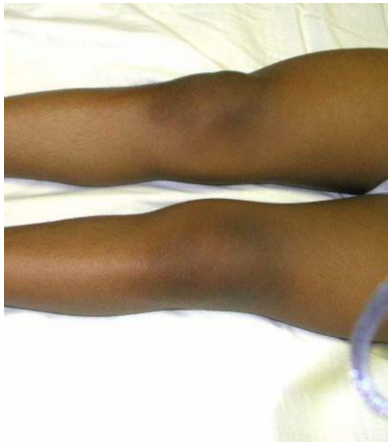
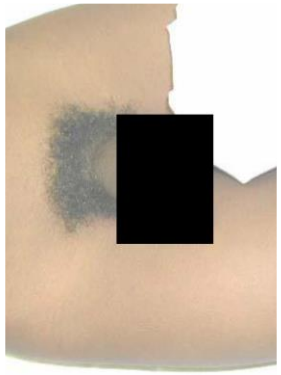
Five -year-old boy previously well, started to develop pubic hair, which has been increasing steadily, adult type body odor, and acne on his back (photos). On examination, pubic hair and penis Tanner stage 2, testicular volumes of 3 ml bilaterally. Which one of the following is most likely diagnosis?

a) Non – classical CAH
b) Premature Adrenarche.
c) Hypothalamic hamartoma.
d) Idiopathic precocious puberty.







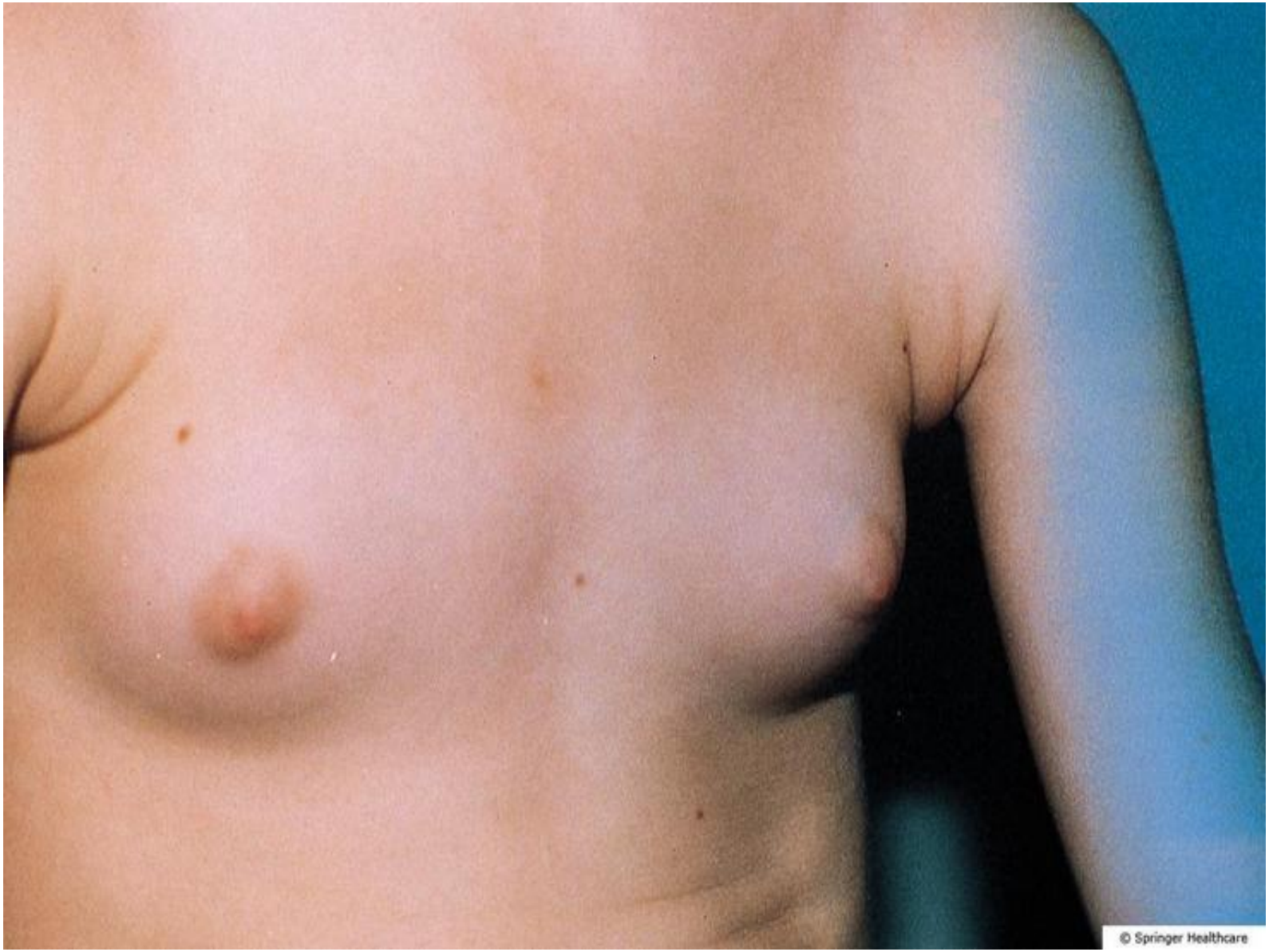
"Non classic" / late-onset CAH

- It does not manifest with neonatal genital ambiguity; rather, it presents later in life with signs of androgen excess.
- Clinical features in late childhood include premature pubarche, acne, and accelerated bone age.
- In adolescent girls and adult women, non classic CYP21A2 deficiency is characterized by acne, hirsutism, and menstrual irregularity (oligoovulation) that are indistinguishable from the polycystic ovary syndrome.
- Never presents with adrenal crisis.

Four -year-old girl presented with bilateral breast enlargement & vaginal discharge, together with moodiness and body odor, no relevant past medical history. She was well with no headaches, visual disturbance or polydipsia. Mother and two elder sisters had early menarche at 10–11 years.

On examination, her height is on the 90th% and mid-parental height 50th%. Tanner stage is of B3, PH2, A1. Which one of the following is most important investigation?

a) Observation of further progression of pubertal signs.
b) Bone age assessment.
c) MRI pituitary to look for CNS tumor.
d) Basal &GnRH stimulation test.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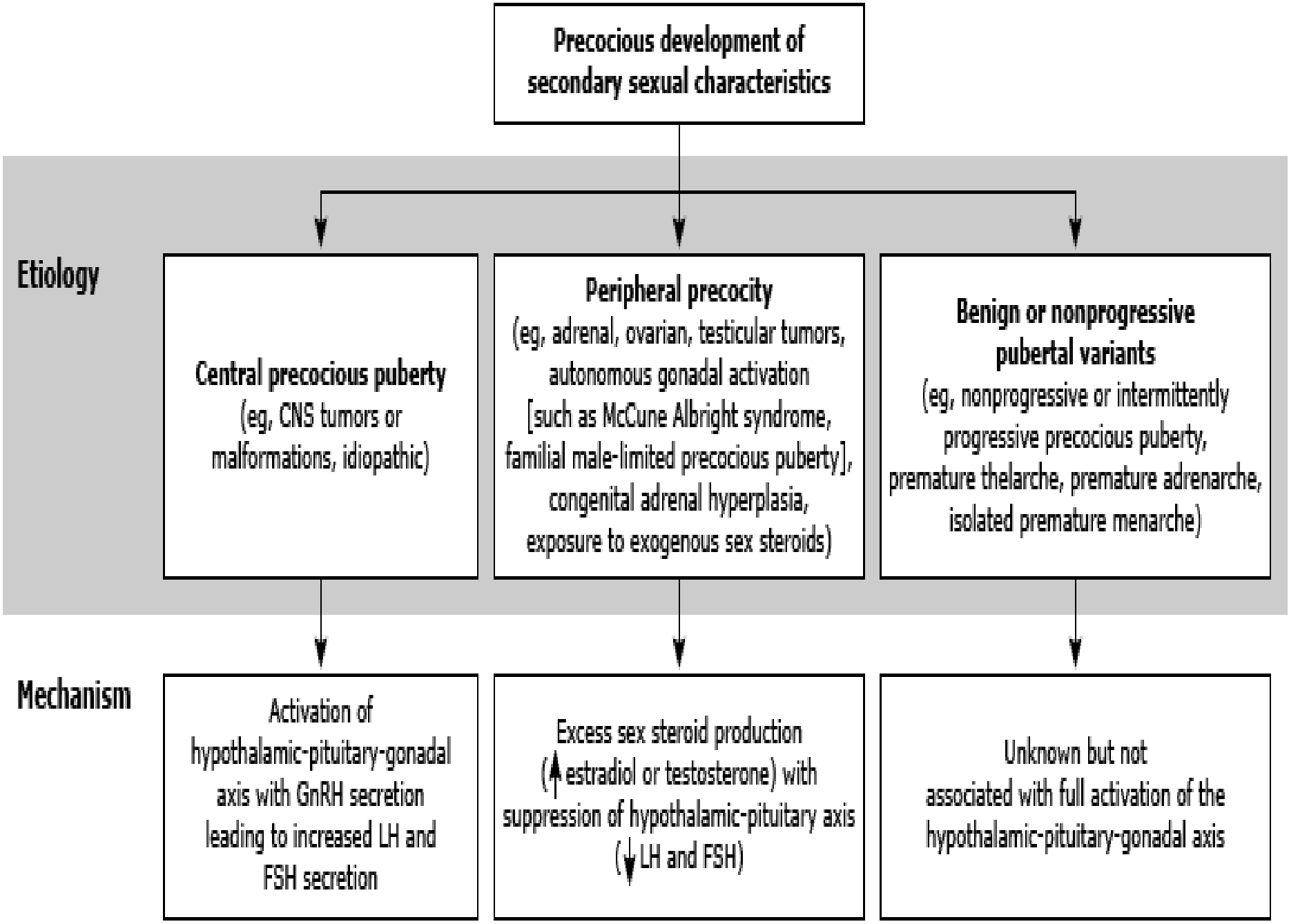
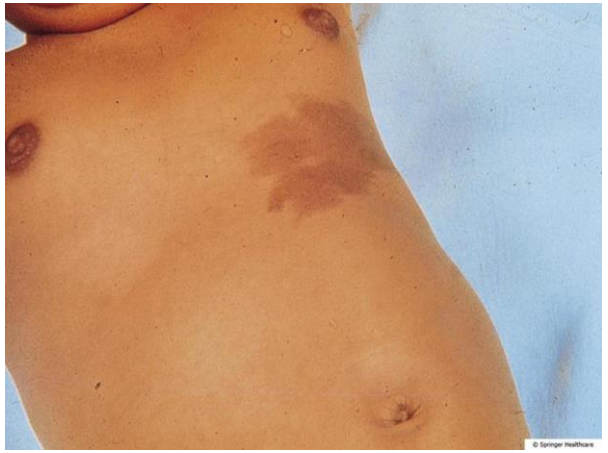
Precocious Puberty

- Central precocious puberty (CPP) is caused by an early activation of the hypothalamic-pituitary-gonadal axis.
  - CPP is pathologic in up to 40 - 75 % of boys & 10 - 20 % of girls.
- Peripheral precocity is caused by:
  - secretion of sex hormones either from the gonads or adrenal glands, ectopic human chorionic gonadotropin (hCG) production by a germ-cell tumor, or by exogenous sources of sex steroids
  - Is independent from the hypothalamic-pituitary-gonadal axis


Precocious development of secondary sexual characteristics
Etiology
Central precocious puberty (eg, CNS tumors or malformations, idiopathic)
Peripheral precocity (eg, adrenal, ovarian, testicular tumors, autonomous gonadal activation [such as McCune Albright syndrome, familial male-limited precocious puberty], congenital adrenal hyperplasia, exposure to exogenous sex steroids)
Benign or nonprogressive pubertal variants (eg, nonprogressive or intermittently progressive precocious puberty, premature thelarche, premature adrenarche, isolated premature menarche)
Mechanism
Activation of hypothalamic-pituitary-gonadal axis with GnRH secretion leading to increased LH and FSH secretion
Excess sex steroid production (↑ estradiol or testosterone) with suppression of hypothalamic-pituitary axis (↓ LH and FSH)
Unknown but not associated with full activation of the hypothalamic-pituitary-gonadal axis

Five- year old girl, brought by her mother because of bilateral breast enlargement and spotty vaginal discharges. On examination (see photo). Her basal pubertal investigations revealed:

- Estradiol 62 pg/ml (<10)
- FSH <0.1 mIU/mL
- LH <0.1 mIU/mL



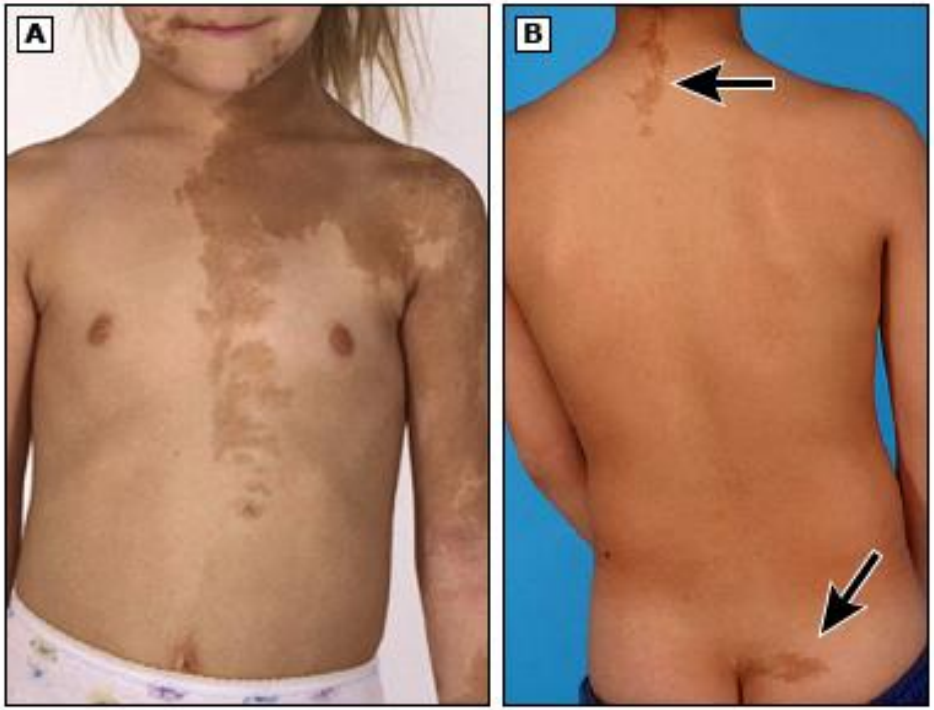
Which one of the following, is the most important confirmatory investigation you will order?

a) GnRH stimulation test.
b) Gene mutation screening.
c) Thyroid function test.
d) Skeletal survey.

McCune-Albright syndrome (MAS) consists of at least 2 of the following 3 features:

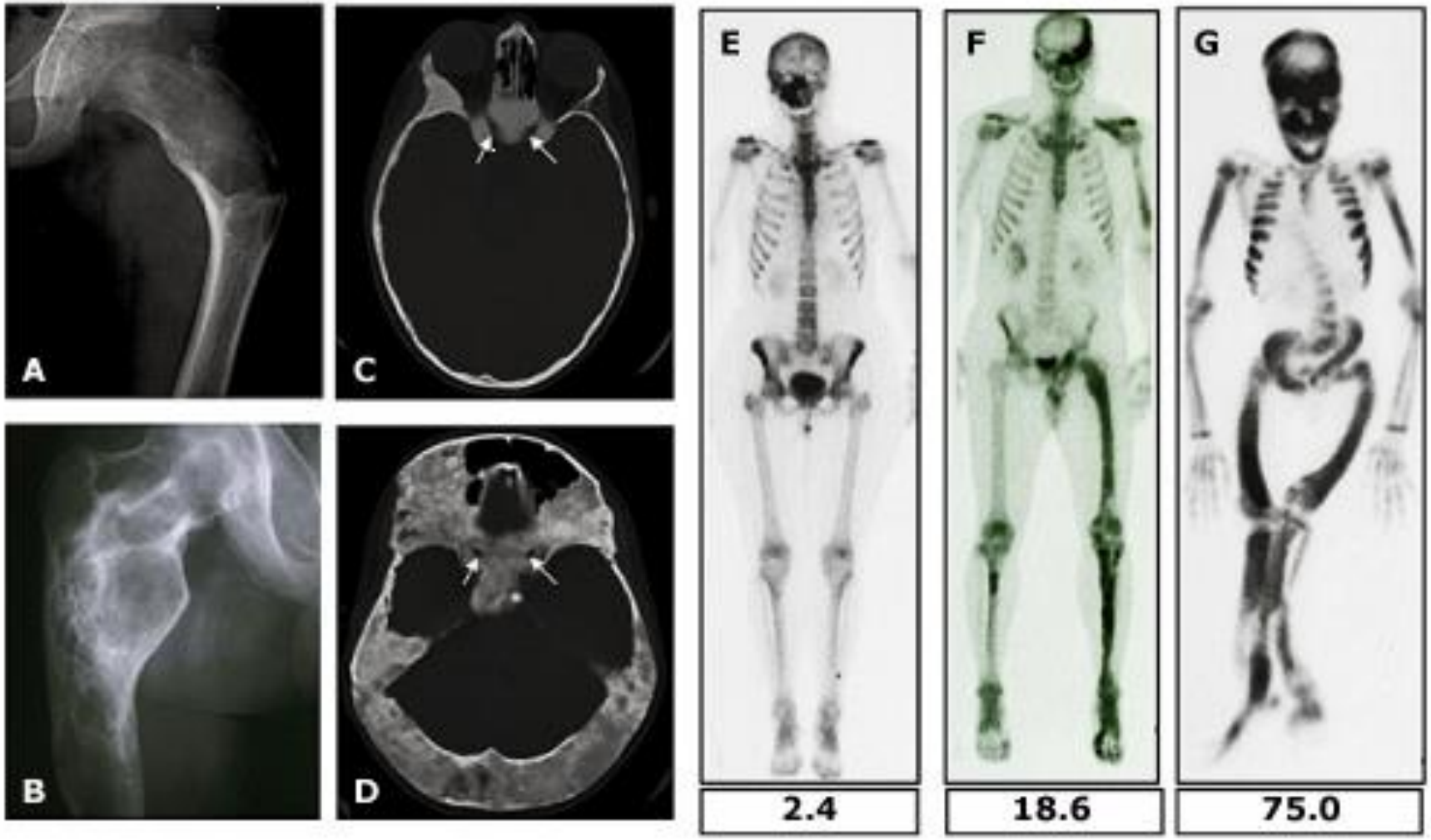
- Polyostotic fibrous dysplasia (PFD).
- Café-au-lait skin pigmentation,
- Autonomous endocrine hyperfunction (e.g., gonadotropin-independent precocious puberty).
- Other endocrinopathies may be present, including hyperthyroidism, acromegaly &Cushing syndrome.
- Genetically, activating mutation of the GNAS1 gene , which is involved in G- protein signaling.

Café-au-lait spots in McCune-Albright syndrome

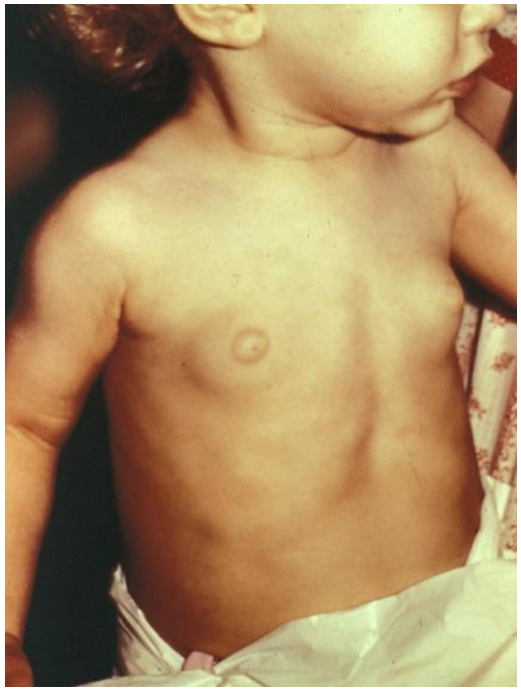


(A) A typical lesion on the face, chest, and arm of a five-year-old girl with McCune-Albright syndrome, which demonstrates jagged "coast of Maine" borders and the tendency for the lesions to both respect the midline and follow the developmental lines of Blaschko (a configuration of skin lesions characterized by arcs on the upper chest, S shapes on the abdomen, and V shapes over the posterior midline, caused by patterns of X-chromosome inactivation).
(B) Typical lesions that are often found on the nape of the neck and crease of the buttocks are shown (arrows).

Radiographic appearance of fibrous dysplasia in McCune-Albright syndrome



Two- year old girl with bilateral breast development with no growth acceleration, no bone age advancement & normal estradiol, LH or FSH. What is the most likely diagnosis ?



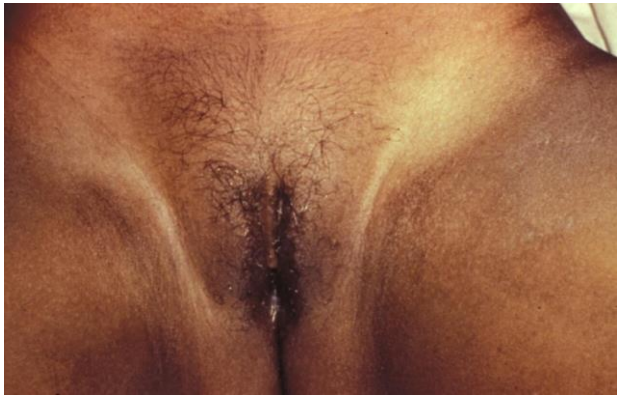
a) Ingestion of her mother's OCPs.
b) Precocious puberty.
c) Benign isolated premature thelarche.
d) McCune Albright Syndrome.

Benign Premature Thelarche

- Isolated breast development
  - 80% before age 2 years.
  - Rarely after age 4 years.
- Not associated with other signs of puberty. (growth acceleration, advancement of bone age)
- Children go on to normal timing of puberty and normal fertility.
- It may be associated with functional follicular cysts that spontaneously regress and perhaps with especially responsive breast tissue
- Benign process.
- Routine follow-up.

Six- year old girl, with 6 months history of pubic hair growth associated with fine axillary hair as well as adult odor to sweat. No breast development with no acceleration of growth. Otherwise normal history and examinations.

What is the most likely diagnosis?

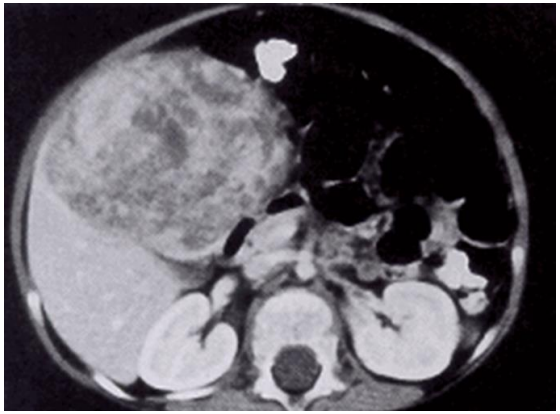


a) Precocious puberty.
b) Benign premature Adrenarche.
c) Non-classical congenital adrenal hyperplasia.
d) Adrenal tumor.

Benign premature adrenarche

- Production of adrenal androgens before true pubertal development begins.
- Presents as isolated pubic hair in mid childhood
  - No growth acceleration.
  - No testicular enlargement in boys.
- If normal growth rate, routine follow-up.
- If accelerated growth and/or bone age advancement, screen for:
  - CAH
  - Virilizing tumor (adrenal/gonadal)

Two - year old girl, presented with abdominal distension, there was three months history of bilateral breast enlargement, pubic hair appearances & rapid growth. Her abdominal CT scan (photo).



Which one of the following is most likely diagnosis?

a) β-HCG secreting hepatoblastoma
b) McCune Albright syndrome.
c) Benign premature Adrenarche.
d) Ovarian cyst.

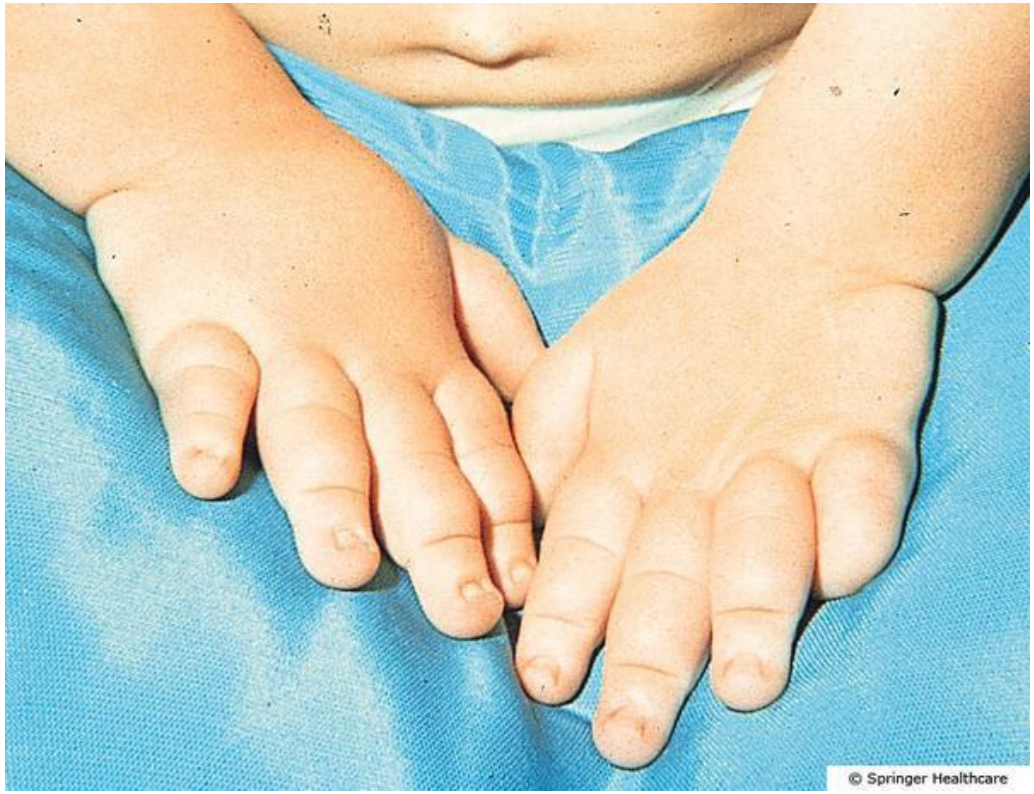
Gonadotropin independent precocious puberty secondary to β-HCG secreting hepatoblastoma

Hepatoblastoma

- Is the most common primary hepatic malignancy in early childhood.
- The majority of cases occur in the first two years of life and rarely in children older than five years.
- Syndromes with an increased incidence of hepatoblastoma include:
  - Beckwith Wiedemann syndrome.
  - trisomy 18 & trisomy 21 .
  - Aicardi syndrome.
  - Li-Fraumeni syndrome.
  - Goldenhar syndrome.
  - type 1a glycogen storage disease.
  - familial adenomatous polyposis.
- Serum alpha-fetoprotein (AFP) levels are markedly elevated.
- Sexual precocity may be present due to the synthesis of ectopic gonadotropin (HCG).

Thirteen-year-old girl, is referred with growth failure with delayed puberty. On examination her height is below the 3rd % . (Please see photo).Which one of the following is most likely diagnosis?

a) Constitutional delay of Puberty.
b) Hypogonadotropic hypogonadism.
c) Hypopituitarism.
d) Hypergonadotropic hypogonadism.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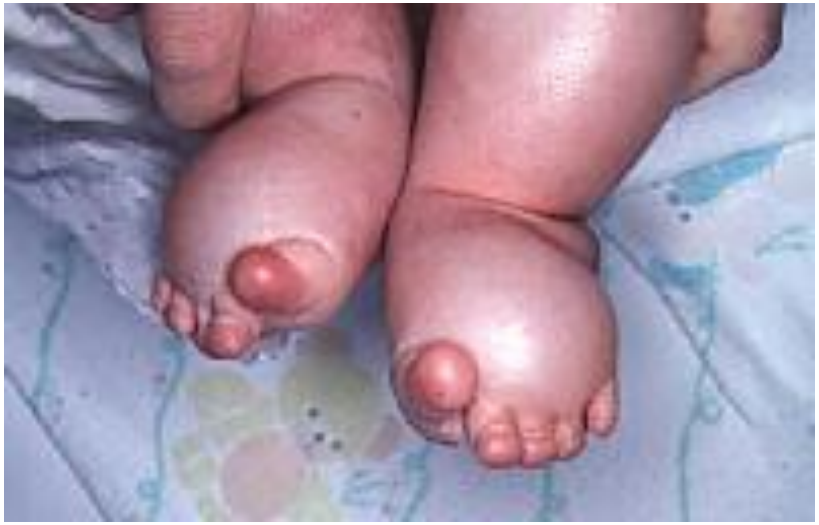
Ovarian failure

- Turner syndrome is one of the most common causes of premature ovarian failure.
- Most affected girls have no breast development and have primary amenorrhea.
- Approximately 15 - 30 % of girls with Turner syndrome either have initial breast development followed by pubertal arrest, or complete puberty but then develop secondary amenorrhea.
- A small percentage of girls have normal pubertal development and regular menstruation.
- These milder phenotypes of ovarian failure are more common in girls with mosaicism compared with those with 45,X monosomy.

Full term neonate, is born with isolated bilateral swollen feet.



What is the most likely diagnosis?

a) Cardiac failure with lower limb edema.
b) Systemic allergic reaction.
c) Congenital nephrotic syndrome.
d) Turner syndrome.

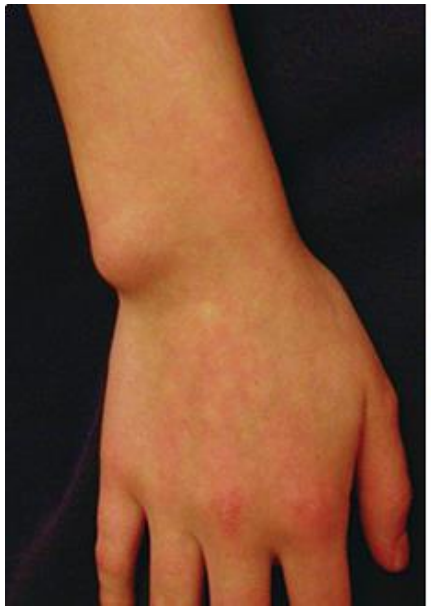
Turner syndrome

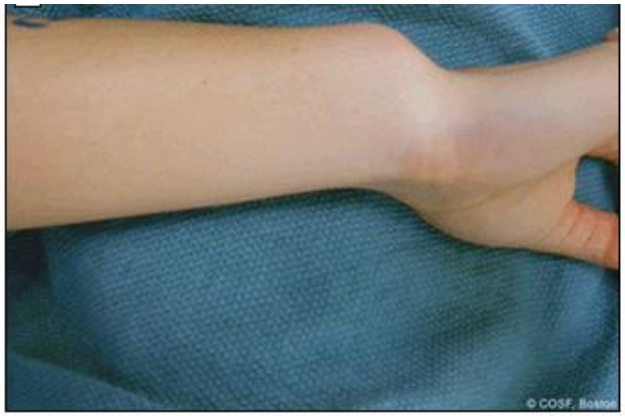
- Combination of short stature, primary amenorrhea (ovarian dysgenesis), webbed neck, lymphedema, and cubitus valgus.
- Incidence among live born female infants of 1: 5000.
- More than half have 45, X karyotype.
- The remainder show mosaicism and/or more complex rearrangements involving the X chromosome.
- Between 20% and 40% of girls with Turner syndrome have significant heart defects, most commonly coarctation of the aorta (70%), often bicuspid aortic valve, and aortic stenosis.

Type and frequency of chromosome abnormalities in Turner syndrome

Karyotype	Percent (%)	Description
45,X	40 to 50	Monosomy X
45,X/46,XX	15 to 25	Monosomy X
45,X/47,XXX; 45,X/46,XX/47,XXX	3	Mosaicism with "Triple X"
45,X/46,XY	10 to 12	Mixed gonadal dysgenesis
46,XX, del(p22.3); 46,X,r(X)/46,XX	10 to 12	Deletion Xp22.3
	10 to 12	Ring X chromosome
46,X i(Xq); 46,X,idic (Xp)	(~10)	Isochromosome Xq; isodicentric Xp
X-autosome translocation, unbalanced	Rare	Various
46,XX,del(q24)	Rare	Not TS; premature ovarian failure
46,X,idic(X)(q24)	Rare	Not TS; isodicentric Xq24

Ten –year - old girl, presented with arm deformity (photo). What is the diagnosis?

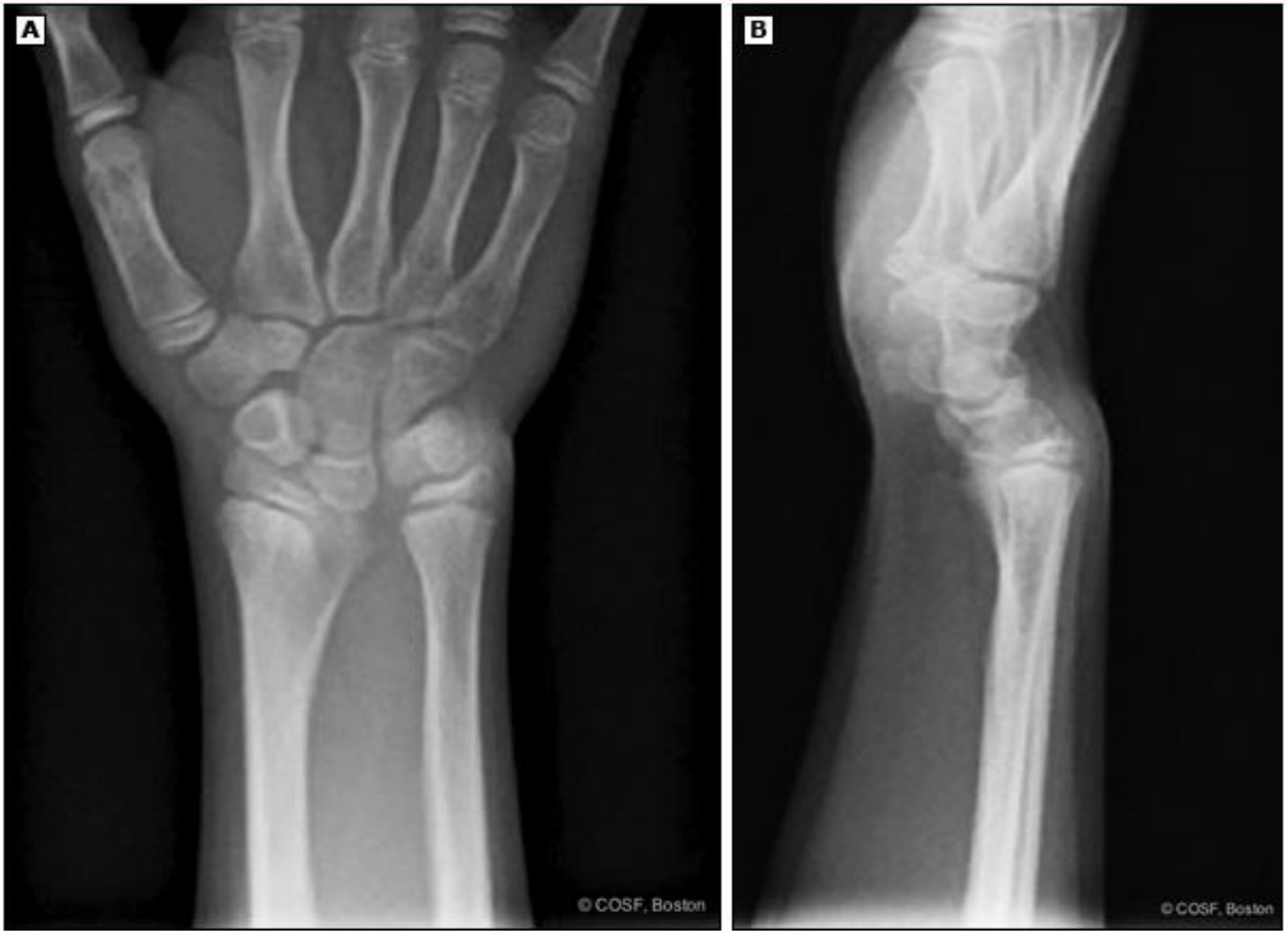




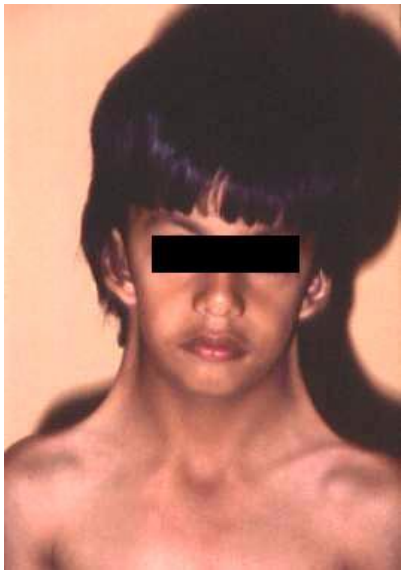
Madelung deformity

- Madelung deformity is a focal dysplasia of the distal radial epiphysis.
- Premature closure leads to a progressive deformity with dorsal displacement of the ulna.
- Madelung deformity occurs in girls with Turner syndrome and is usually bilateral.
- The diagnosis of Madelung deformity is made radiographically; radiographs of both wrists, forearms, and elbows should be obtained.

Madelung deformity radiographs



Seven-year -old boy has presented to the endocrinology clinic with short stature & mental subnormality. What is the most likely diagnosis?



a) Turner's syndrome.
b) VATER syndrome.
c) Silver – Russel syndrome.
d) Noonan's syndrome.

Noonan syndrome (NS)

- Autosomal dominant disorder.
- The classical features include:
  - short stature.
  - congenital heart disease (CHD).
  - clinical features of NS include difficulties with feeding in early life; vision, hearing, and growth problems; specific learning difficulties & easy bruising and bleeding.

Ten- year female presented, with a 4 week history of polyuria, polydipsia, and unexplained weight loss. She was noticed to have deep, sighing respiration. Glucose was 498 mg/dL, pH 7.06. Which one of the following is the most important initial management ?

a) Insulin drip 0.1 units/kg/hour.

b) ½ Normal saline with 40 mmol of potassium at twice maintenance.

c) Bicarbonate 1 mmol/kg slowly over 1 hour.

d) Fluid rehydration with 0.9 NS.

Sixteen -year old boy, presented with delayed puberty. He was having a recurrent episodes of headache, diplopia and increased urination. His height was < 3rd percentile. Which one of the following is the most likely diagnosis?

a) Diabetes mellitus.
b) Cerebellar tumor.
c) Craniopharyngioma.
d) Kalman's syndrome.

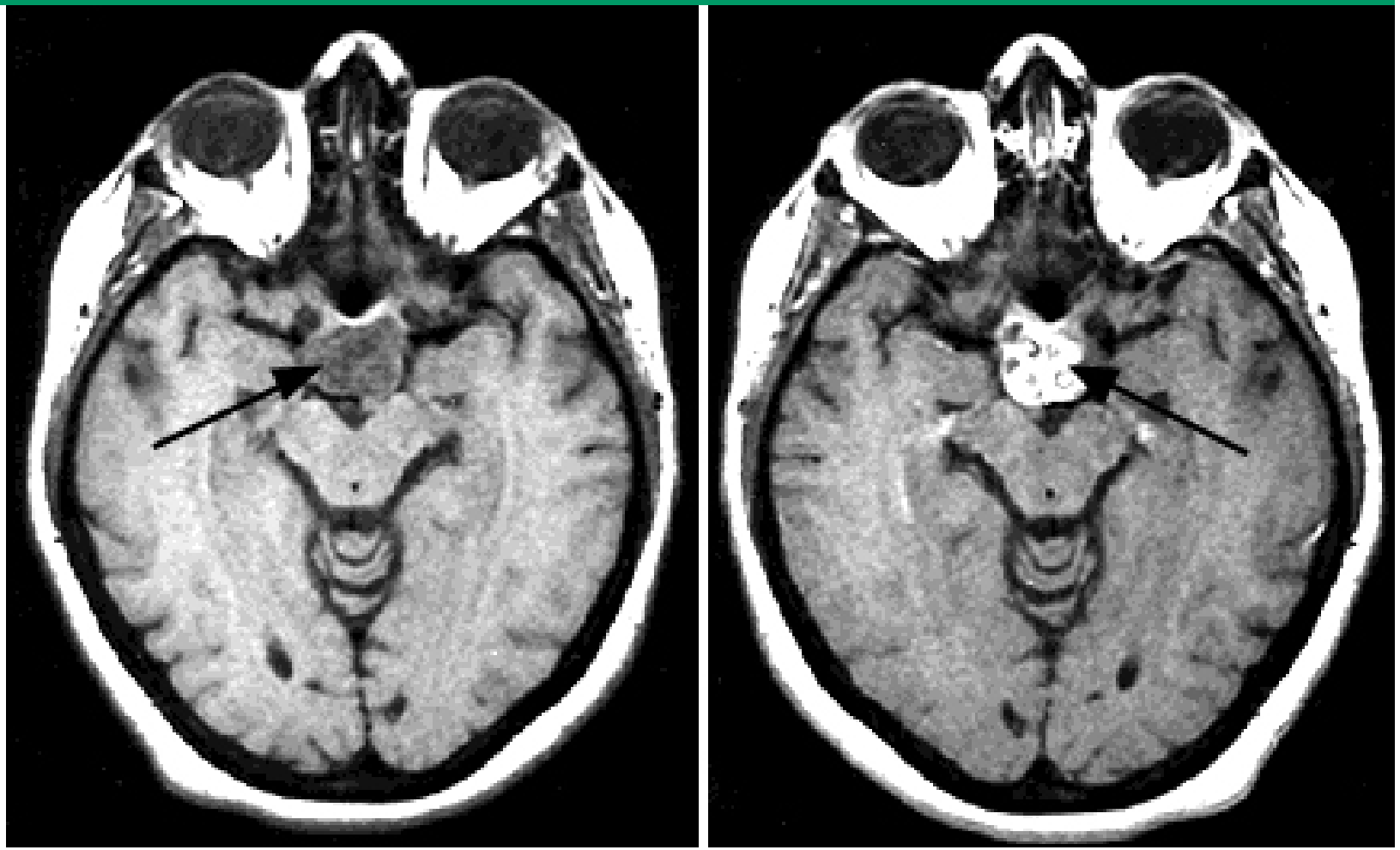
Craniopharyngioma

- Benign tumor with tendency to recur after excision and the high surgical risk due to involvement of the most vital structures of the brain.
- Rare solid or mixed solid & cystic tumors that arise from remnants of Rathke's pouch.
- Historically, been named as "Rathke pouch tumors“.
- A wide range of symptoms may be present.
- Visual symptoms are frequent, result from compression of the optic chiasm or nerves.
- Moderate to severe daily headaches are present in approximately 50 % of patients at the time of diagnosis.
- Other generalized symptoms, such as depression, nausea, vomiting, and lethargy can accompany pressure-related headaches.

Craniopharyngioma

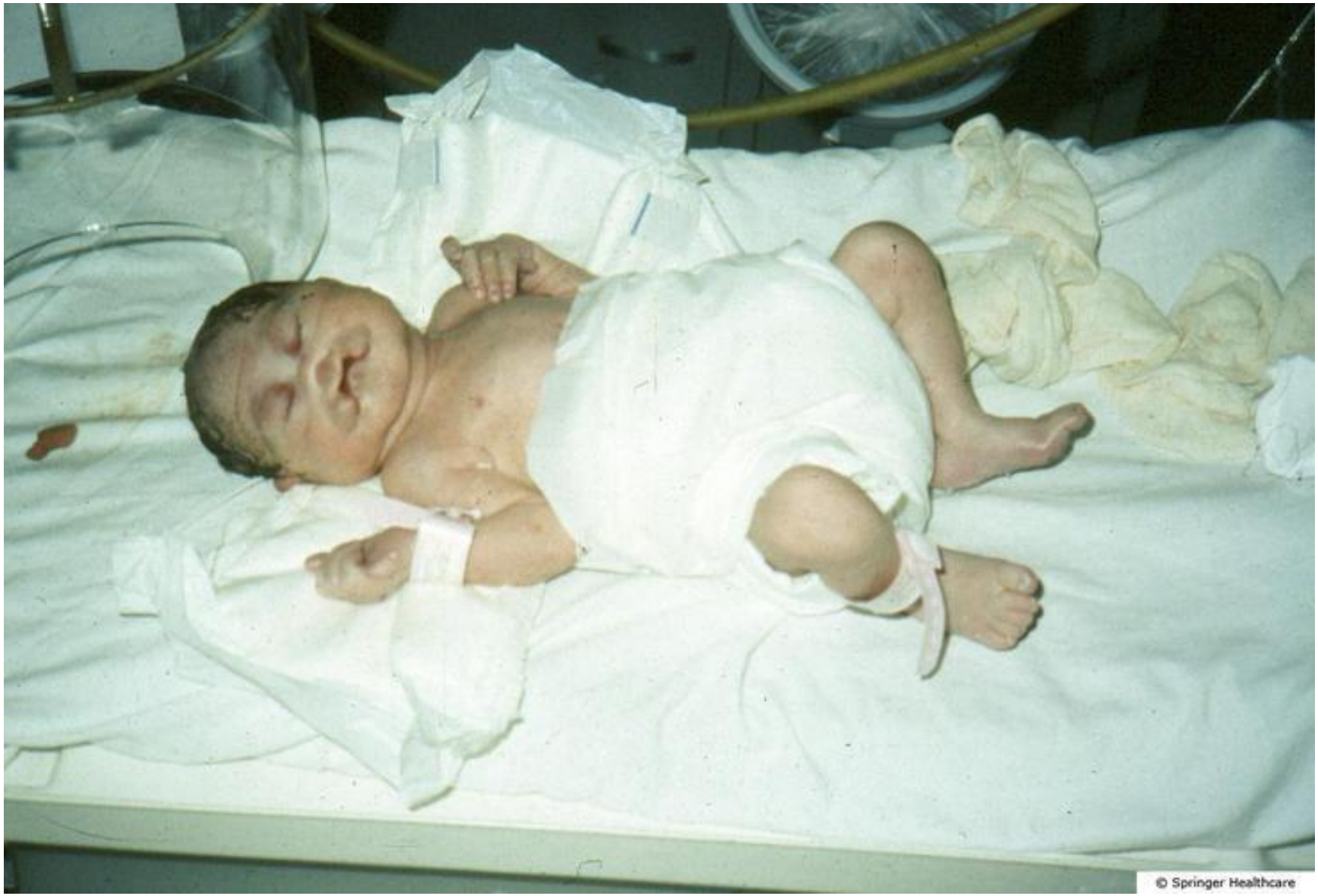
- Endocrine abnormalities due to direct damage to or compression of normal structures or post surgical removal of the tumor can lead to a range of endocrine abnormalities.
- Frequently observed complications include diabetes insipidus (75%).
- Growth failure, caused by either hypothyroidism or growth hormone deficiency, is the most common presentation in children.
- Delayed puberty could be seen in 40 % of cases.

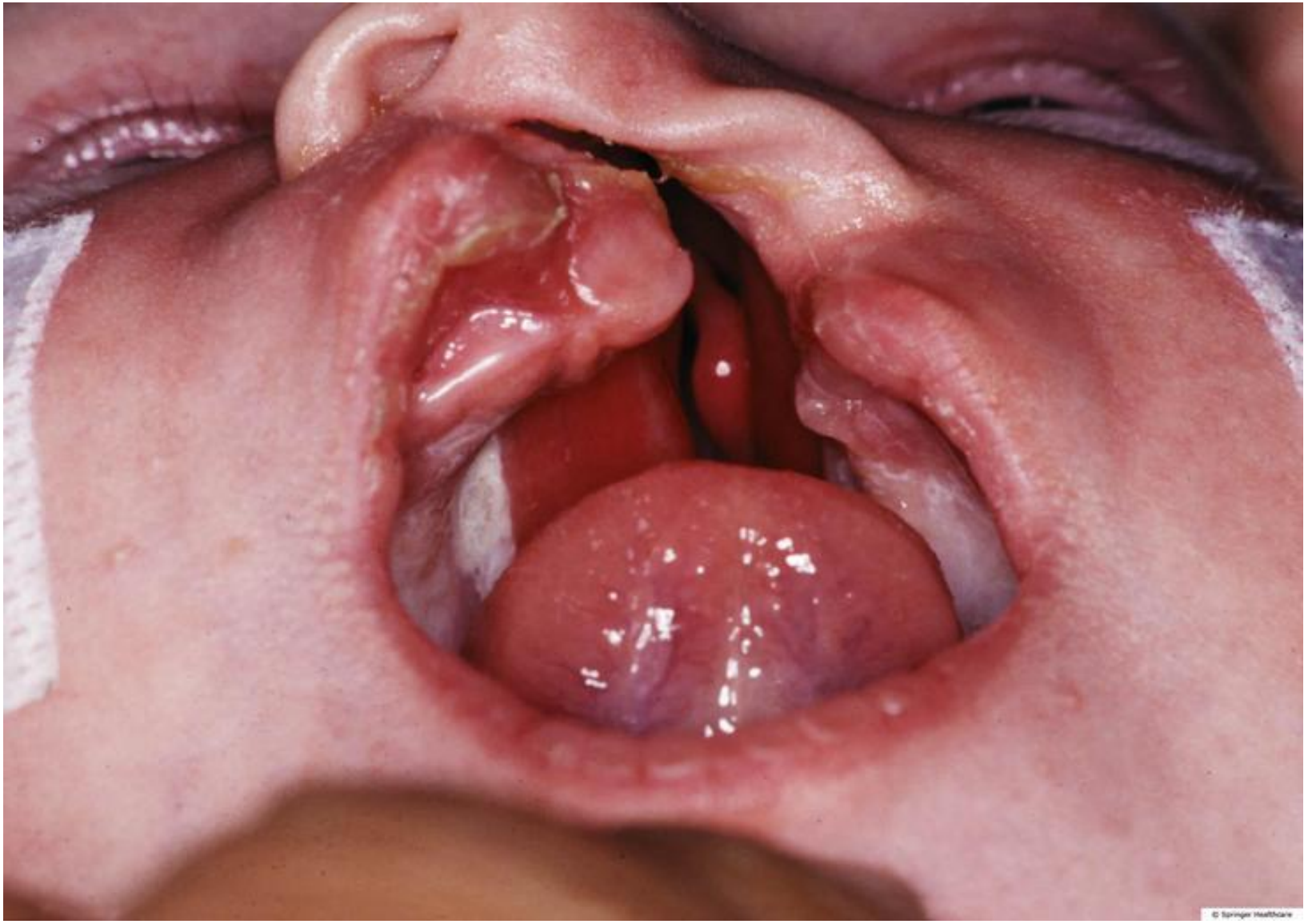
Craniopharyngioma



Twenty – day- old, neonate was seen in the pediatric endocrinology clinic for recurrent episodes of hypoglycemia . On examination, he had a cleft lip and palate with a small mid-face. Both testes were palpable, however short penile length was measured. What is your best approach in order to reach to the final cause of hypoglycemia?

a) Look for other dysmorphic features.
b) Admit to do critical samples during his hypoglycemia attack.
c) Do GH stimulation test.
d) Do MRI brain.

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Holoprosencephaly (HPE)

- Is cephalic disorder in which the forebrain of the embryo fails to develop into two hemispheres.
- Normally, the forebrain is formed and the face begins to develop in the fifth and sixth weeks of gestation.
- The condition can be mild or severe.
- Most cases are not compatible with life and result in fetal death in-utero.
- When the embryo's forebrain does not divide to form bilateral cerebral hemispheres (the left and right halves of the brain), it causes defects in the development of the face , brain & pituitary structure and function.

Hypopituitarism

- Affects between 1 in 4,000 -10,000 live births, with increasing incidence with age.
- Congenital hypopituitarism most often results from genetic or embryologic pathologies.
- The clinical presentation of hypopituitarism, widely varies, depends on the patient's age, the etiology, and the specific hormone deficiencies, which may occur as isolated deficiencies or in various combinations of MPHD.

Hypopituitarism

- Most neonates with hypopituitarism have normal birth weights and lengths and no history of intrauterine growth retardation.
- However, they often have histories of breech presentation (particularly neonates with MPHD), although the explanation for this is unclear.
- The hypoglycemia risk is higher in neonates with hypopituitarism, with various manifesting symptoms, such as lethargy, jitteriness, pallor, cyanosis, apnea, or convulsions.
- Jaundice may be secondary to indirect hyperbilirubinemia (TSH deficiency) or to direct hyperbilirubinemia (GH or ACTH deficiencies).

GOOD LUCK

End of Part 1
MCQs Revision