Pediatric Endocrinology Review MCQs

PART - 5

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Seven - year old girl, presented to the endocrine clinic because of short stature, which was reported since birth. She continued to be shorter than her schoolmates. On examination (photo). Which one of the following is a diagnostic investigation?

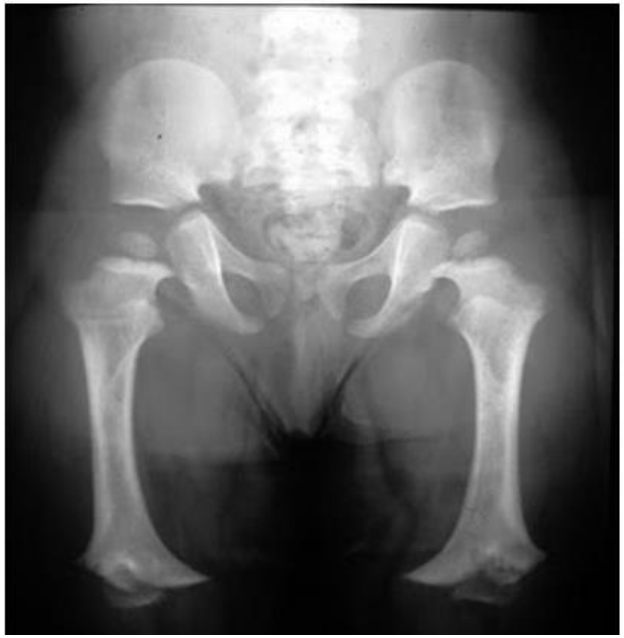
a) Bone age assessment.
b) Skeletal survey.
c) Thyroid function test.
d) Chromosomal analysis.

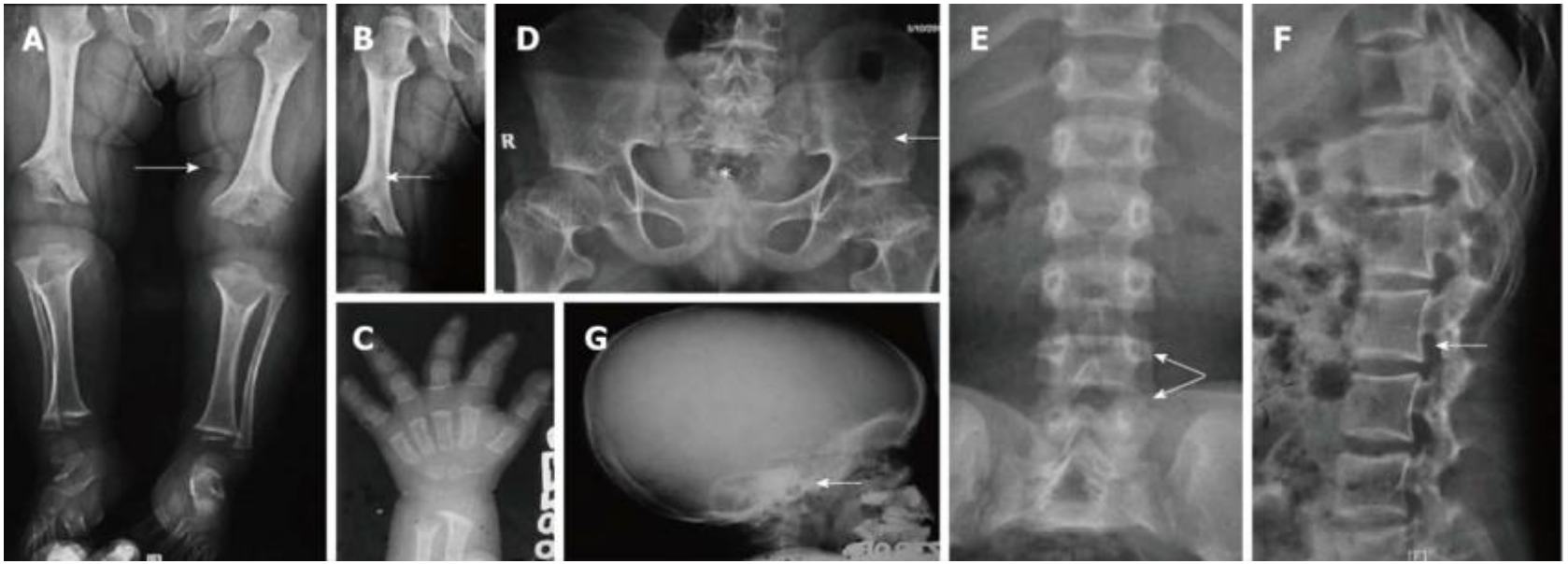
Achondroplasia

- Occurs due to sporadic mutations in the majority of cases but can be inherited as autosomal dominant condition.
- Achondroplasia is the most common form of disproportionate short stature.
- Generally recognizable intrauterine because of short limbs & macrocephaly.
- Limb shortening is predominantly in the proximal segment (rhizomelic).
- A **trident hand** is a description where the hands are short with stubby fingers, with a separation between the middle and ring fingers.
- Children with achondroplasia have normal intelligence.
- Specialized growth charts have been developed for head circumference, height.
- They have significant midface hypoplasia, which increases the risk of obstructive sleep apnea.
- The narrowing of the foramen magnum can cause brainstem compression with an increased incidence of sudden infant death.

Achondroplasia

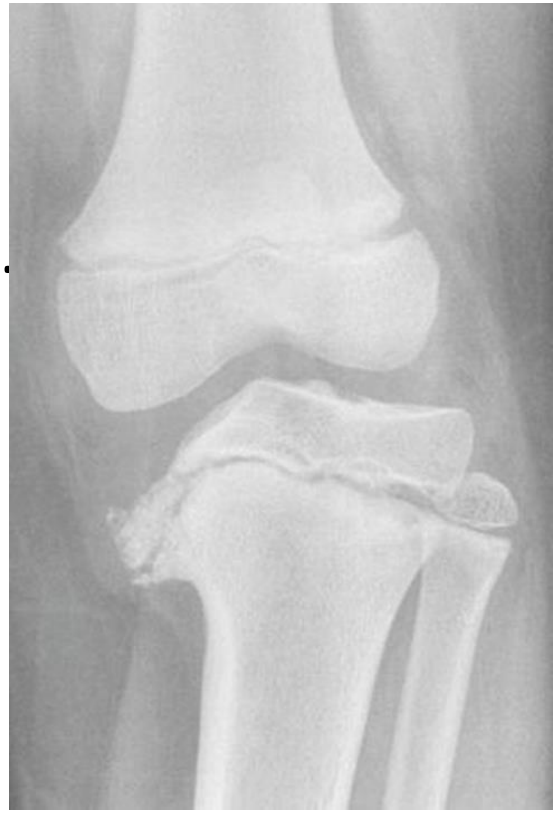
- Pelvis is abnormal with small, square iliac wings
- Horizontal acetabular roots and narrowing of the greater sciatic notch
- long bones are short and the metaphyses slope
- because of narrow chest - respiratory problems are frequent
- translucent area at proximal ends of the femora in neonatal period

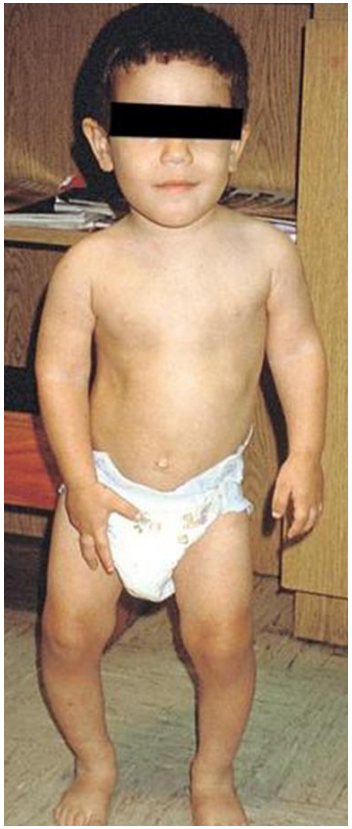




Eighteen – months old boy, brought by his parents because of increasing bowing of his legs (photo). Nutritional history was unremarkable. He was on vitamin D prophylactic daily dose (400 units) since birth. His limb x-ray (photo). Which one of the following is most likely diagnosis?

a) Osteodystrophy.
b) Vitamin D resistant rickets.
c) Blount's disease.
d) Hypophosphatasia.

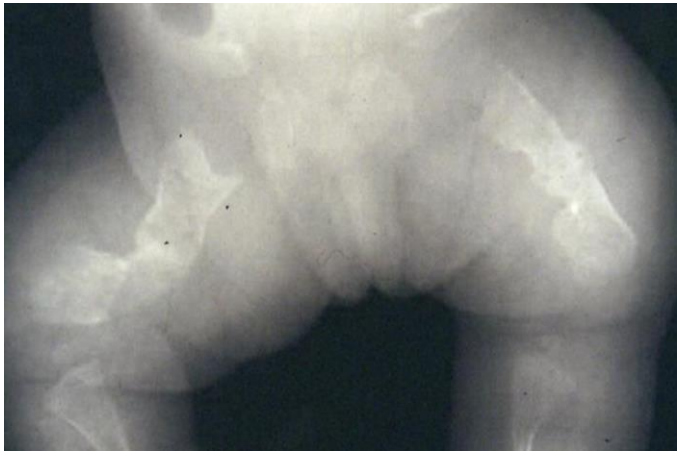


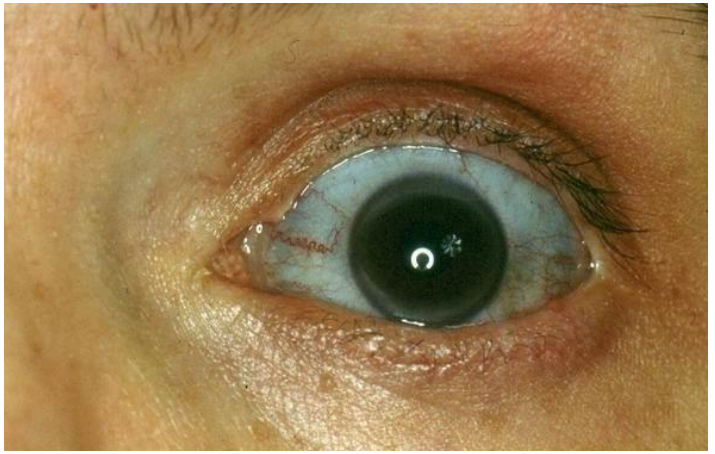


Tibia vara (Blount’s disease)

- Is a growth disorder of the tibia that causes tibial bone to angle inward, resembling a bowleg.
- It is also known as "tibia vara".
- Described by Dr. Walter Blount (1900–1992) “American pediatric orthopedic surgeon”.
- Blount's disease occurs in young children & adolescents.
- The cause in majority of cases is unknown.
- Sometimes associated with obesity due to the effects of weight on the growth plate.
- Is progressive disease.
- Severe bowing of the legs “unilateral or bilateral”.

Eight -week-old infant, brought by his mother because of previous fractures of the left femur & right tibia happened during labor. Mother noticed some eye change (photo). Plain x-ray of his lower limb (photo). What is most likely diagnosis?



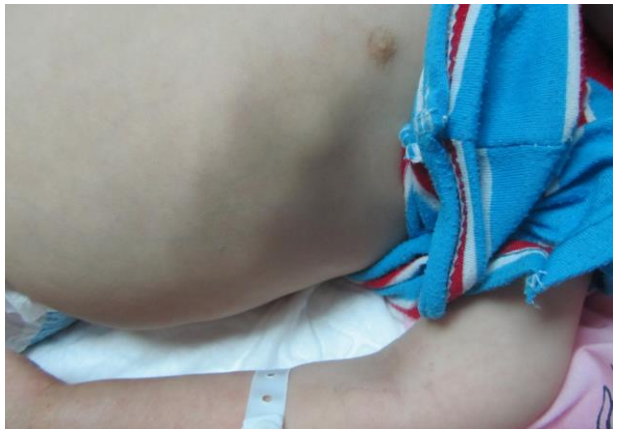


a) Hypophosphatasia.
b) Osteogenesis imperfecta.
c) Congenital vitamin D deficiency.
d) Hypophosphatemic rickets.

Osteogenesis imperfecta (OI)

- Characterized by osteoporosis and recurrent fractures “brittle bone disease.”
- Clinically has many subtypes that vary in both the degree of bone fragility and features.
- Blue sclera is present in some types(which can be a normal variant in the neonatal period) and dentinogenesis imperfecta.
- Type II is lethal type and is often identified prenatally by ultrasound.
- Type III is severe but not lethal, and generally the affected children are born with numerous fractures and bowed limbs.
- Types I & IV less severe but more common forms of OI, and patients usually have normal stature but may have multiple fractures in childhood.
- Associated features include easy bruising & joint hypermobility.

Two-year-old boy was referred for further assessment of his increasingly bow legs. His maternal uncle has rickets. On chest examination (photo). blood measurements were obtained: calcium 2.37mmol/l, phosphate 0.13mmol/l , alkaline phosphatase 805IU/l, PTH 1.3pmol/l. Which one of the following statement is true?

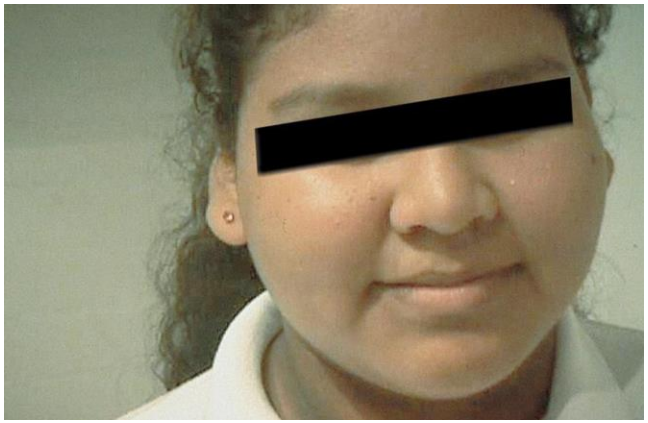


a) Nutritional rickets.
b) Vitamin D dependent type 1 rickets.
c) Vitamin D dependent type 2.
d) X- linked hypophosphatemic Rickets.

X-linked hypophosphatemic Rickets (XLH)

- Hereditary rickets.
- Is associated with PHEX gene mutation.
- The PHEX gene regulates fibroblast growth factor 23 (FGF23).
- FGF 23 normally inhibits renal reabsorption of phosphate into the bloodstream.
- Serum calcium within normal or slightly below the reference range.
- Serum phosphate low, alkaline phosphatase high, parathyroid hormone normal.
- Most importantly, urinary loss of phosphate is above the reference range.
- Treatment with oral phosphate & one alpha or calcitriol.
- If severe bowing, an osteotomy performed to correct the leg deformity.
- Burosumab (monoclonal antibody) was licensed in 2018 as the first drug for this condition.

Twelve- year - old obese girl, mother has noticed abnormal discoloration around her neck (photo). Her blood pressure was 155/ 110 (repeated). On investigations, her fasting glucose was 155 mg/dl, her liver enzymes were elevated and her serum triglyceride was high. Which one of the following is most likely diagnosis?



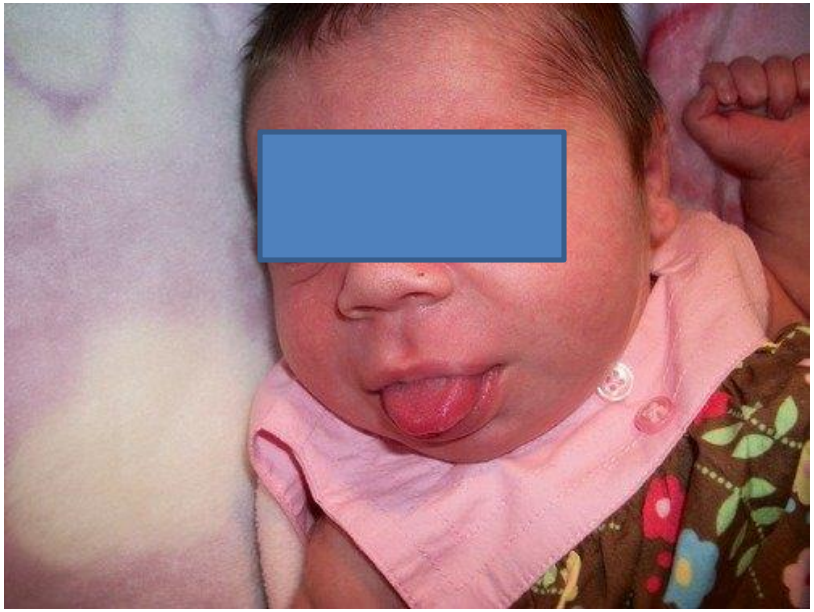
a) Type 2 diabetes.
b) Mody diabetes.
c) Type 1 diabetes.
d) Metabolic syndrome.

Metabolic syndrome in children

- In recent years, greater concern about the presence of obesity & metabolic syndrome in children and adolescents.
- Since 1995, a number of reports of type 2 diabetes occurring in obese children.
- In 1988, Reaven described the relationship of insulin resistance to the development of three associated disorders:
  - type 2 diabetes mellitus, hypertension and dyslipidemia (in releation to increase waist circumference.

Criteria/components	Age		
	6 to <10 years-old	10 to 16 years-old	>16 years-old
Adiposity definition	WC ≥ 90th percentile	WC ≥ 90th percentile	WC ≥ 90 cm (boys) or ≥ 80 cm (girls)
Glucose metabolism	Without cut-off definition for MS diagnosis	Fasting blood glucose ≥ 100 mg/dl	Fasting blood glucose ≥ 100 mg/dl
Dyslipidemia	Without cut-off definition for MS diagnosis	Tg ≥ 150 mg/dl or HDL-ch ≥ 40 mg/dl or taking LLD	Tg ≥ 150 mg/dl or HDL-ch ≥ 40 (boys) or ≥ 50 mg/dl (girls) or taking LLD
Arterial hypertension	Without cut-off definition for MS diagnosis	DBP ≥ 130 or SBP ≥ 85 mmHg or taking AHD	DBP ≥ 130 or SBP ≥ 85 mmHg or taking AHD

Four – day old boy, with birth weight of 4.5 kg, with normal antenatal & natal histories. He developed seizure because of repeated hypoglycemic attacks (lowest RBS reading was 32 mg/dl). His general look (photo). What is the most likely cause of hypoglycemia?

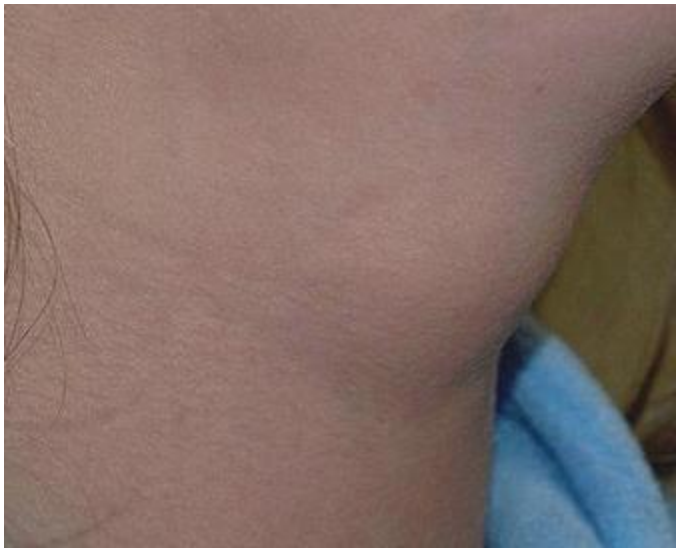


a) Large for gestation age.
b) Infant of diabetic mother.
c) Septicemia.
d) Beckwith-Wiedemann syndrome.

Beckwith-Wiedemann syndrome (BWS)

- BWS is an overgrowth syndrome.
- Is genetically heterogeneous disorder that involves an imprinted region of chromosome 11p15.
- Characterized by:
  - Antenatal & postnatal overgrowth.
  - Macroglossia.
  - Hypoglycemia.
  - Hemihypertrophy.
  - Ear creases or pits
  - Abdominal wall defects (omphalocele).
  - Increased risk of embryonal tumors (Wilms'tumor & hepatoblastoma).
  - Mental retardation is uncommon and usually related to early hypoglycemia.

Six- year old girl, brought to the endocrine clinic because mother has noticed neck swelling. He is otherwise healthy, well developed child. Neck swelling was not painful but increasing in size with growth (photo). Which one of the following is the most likely diagnosis?

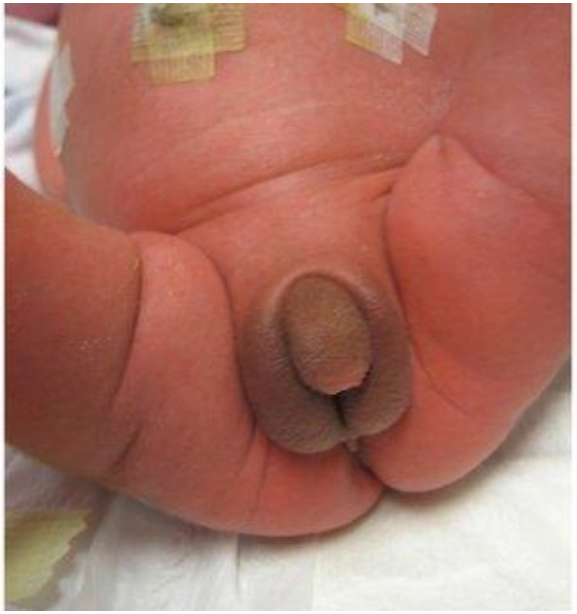


a) Enlarged submandibular lymph node.
b) Small cystic hygroma.
c) Thyroglossal cyst.
d) Laryngocele.

Thyroglossal cyst

- The cyst is painless, soft, round lump in the front of the neck.
- Typically moves, when the child swallows or sticks their tongue out.
- They are often diagnosed in preschool-aged children or during mid-adolescence.
- During fetal development, thyroid gland is located at the back of the tongue which naturally migrates down the neck, passing through the hyoid bone.
- As the thyroid gland descends, it forms a small channel called the thyroglossal duct.
- This duct usually disappears once the thyroid gland reaches its final position in the neck.
- Sometimes part of the duct remains and leaves a pocket.
- A thyroglossal cyst will form when fluid collects in this pocket.
- Surgery should involve removing the entire embryologic remnant.

A neonate was born with abnormal looking genitalia (photo). Chromosomal analysis revealed 46 XX. Which one of the following is most likely diagnosis?

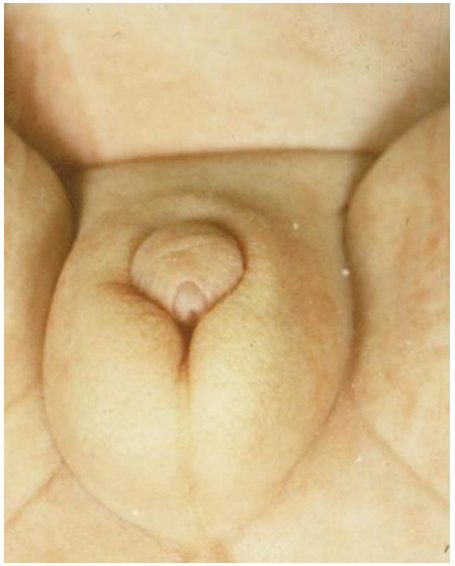


a) Congenital adrenal hyperplasia.
b) Congenital lipoid adrenal hyperplasia.
c) Congenital adrenal hypoplasia.
d) Isolated clitoromegaly.

Congenital adrenal hyperplasia (CAH)

- Autosomal recessive disorder.
- The most common enzyme deficiency of congenital adrenal hyperplasia is 21-hydroxylase deficiency.
- In the severe form associated with mineralocorticoid & glucocorticoid deficiencies.
- Affected female neonates, usually born with ambiguous genitalia.
- Affected male neonates, have normal external genitalia and may present at 2 - 3 weeks of age with weight loss and poor feeding due to adrenal insufficiency.
- To prevent the morbidity and mortality of congenital adrenal hyperplasia, newborn screening programs have been implemented.
- Approximately 75% of patients with CAH due to 21-hydroxylase deficiency have a relative deficiency of mineralocorticoid.

Three weeks old infant, was born at home, brought by his parents because of abnormal looking genitalia (photo). Mother reported that one of her brothers is infertile. Chromosomal analysis revealed 46 XY. Serum testosterone and dihydrotestosterone levels were high. Which one of the following is most likely diagnosis?

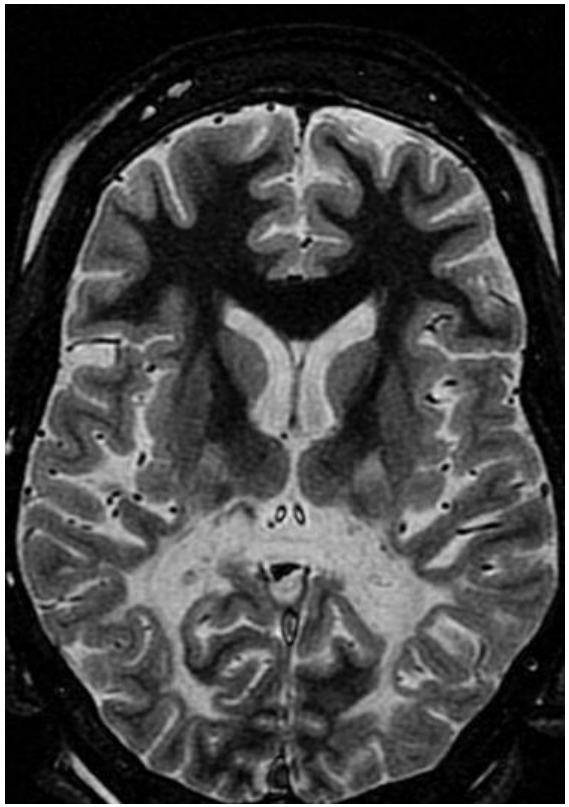


a) Complete androgen insensitivity.
b) Partial androgen insensitivity.
c) 5- α- reductase enzyme deficiency.
d) Testicular biosynthesis defect.

Partial androgen insensitivity syndrome (PAIS)

- The diagnosis is established in an individual with a 46,XY who has:
  - under masculinization of external genitalia.
  - impaired spermatogenesis (adulthood).
  - Normal size & function of both testes.
  - evidence of normal or increased synthesis of testosterone and its normal conversion to dihydrotestosterone.
- AIS represents a spectrum of defects in androgen action and can be subdivided into three broad phenotypes:
  - Complete androgen insensitivity syndrome (CAIS), with typical female external genitalia.
  - Partial androgen insensitivity syndrome (PAIS) with predominantly female, or predominantly male external genitalia.
  - Mild androgen insensitivity syndrome (MAIS) with typical male external genitalia.

Four -year old boy, wo was diagnosed sine neonatal period with primary adrenal insufficiency. Sine then, he was on hydrocortisone replacement therapy with good compliance from his family. Mother has reported that, for the last 6 months, he started to have on/off seizure accompanied with regression of his motor milestone development. He was seen by neurologist who has ordered MRI brain (photo).
Which one of the following is important diagnostic investigation you are going to order?

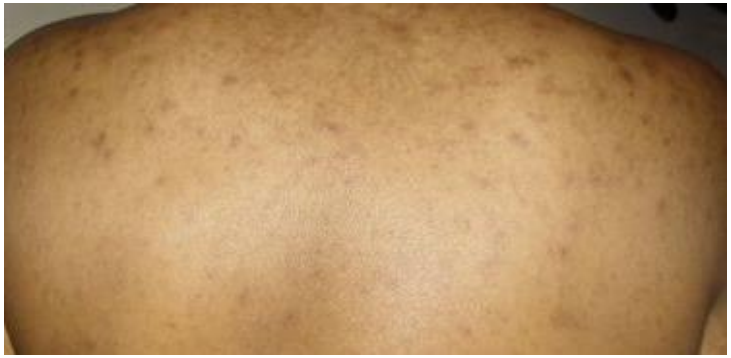


a) Do serum ACTH & cortisol level.
b) Do 17 hydroxyprogesterone level.
c) Do serum electrolytes.
d) Do Very long chai fatty acid (VLCFA).

Adrenoleukodystrophy (ALD)

- ALD is one of a group of disorders caused by a defect of peroxisomes, which are essential for the breakdown of fatty acids in cells.
- ALD mostly affects boys because the disease-causing mutation is located on the X chromosome.
- The condition results in accumulation of very-long-chain fatty acids in nervous system, adrenal gland, and testes.
- There are three major categories of disease:
  - Childhood cerebral form -- appears in mid-childhood (at ages 4 to 8 years)
  - Adrenomyelopathy -- occurs in men in their 20s or later in life.
  - Impaired adrenal gland function (Addison-like phenotype) .

sixteen- year-old adolescent male, was presented to the clinic because of severe anorexia and weight loss for the last 3 months. His back examination (photo). Vital signs were: BP 94/56, HR 105/min. He was dehydrated. His serum glucose random was 55 mg/dl. Which one of the following is important diagnostic investigation to order?



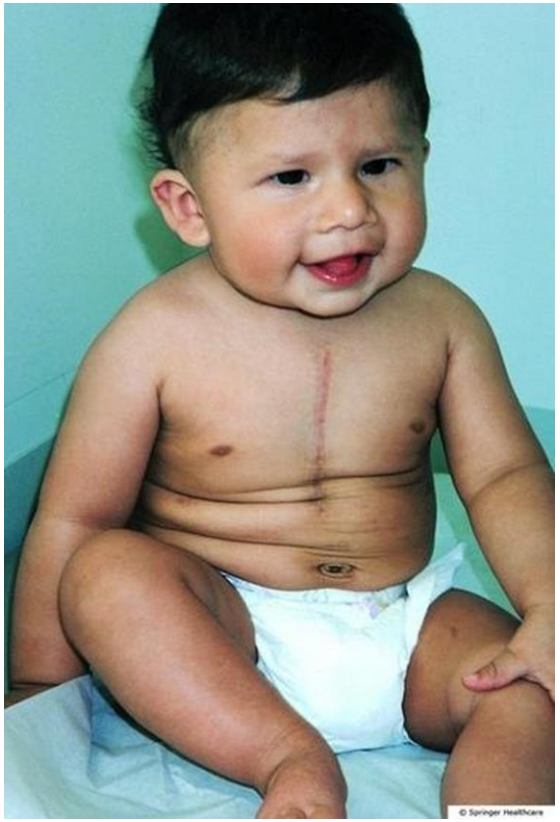
a) Serum ACTH.
b) Serum cortisol & ACTH.
c) Adrenal ultrasound.
d) Serum Renin & Aldosterone.

Addison disease

- Acquired primary adrenal insufficiency results from autoimmune destruction of the adrenal cortex.
- The presenting complaints include: malaise, weakness, failure to thrive, weight loss, anorexia, hypoglycemia, salt craving, abdominal pain, vomiting & hyperpigmentation.
- On examination, the patient may be hypotensive and frequently has increased pigmentation over the knuckles, elbows, knees, ankles, and gums.

Nine - month old boy, was brought by his parents due to repeated attacks of hypocalcemia since birth, with past history of open heart surgery at neonatal period. His general examination (photo).

Which one of the following investigations is essential to reach the diagnosis of hypocalcemia?



a) Serum ionized calcium.
b) Serum phosphate.
c) Serum 25- hydroxy vitamin D.
d) Serum Parathyroid hormone.

DiGeorge syndrome, “22q11.2 deletion syndrome”

- Is caused by deletion of a small segment of chromosome 22.
- The major features include: congenital heart disease, hypocalcemia due to hypoparathyroidism & defective T-cell immunity.
- Truncus arteriosus or interrupted aortic arch are common.
- Facial features include: low-set ears, microstomia, & hypertelorism.
- Embryologic development defects of the third & fourth brachial arches and their derivatives, which includes the parathyroid glands, aortic arch, and thymus gland.
- Infants with low T-cell functions are at risk for common pathogens including candida and herpes simplex, and opportunistic infections, such as *Pneumocystis carinii*.
- The diagnosis can be confirmed by absence of thymus as detected by chest radiography or by direct inspection during surgery to correct their congenital heart defect.

GOOD LUCK

End of part- 5