Pediatric Endocrinology Review MCQs

Part 2

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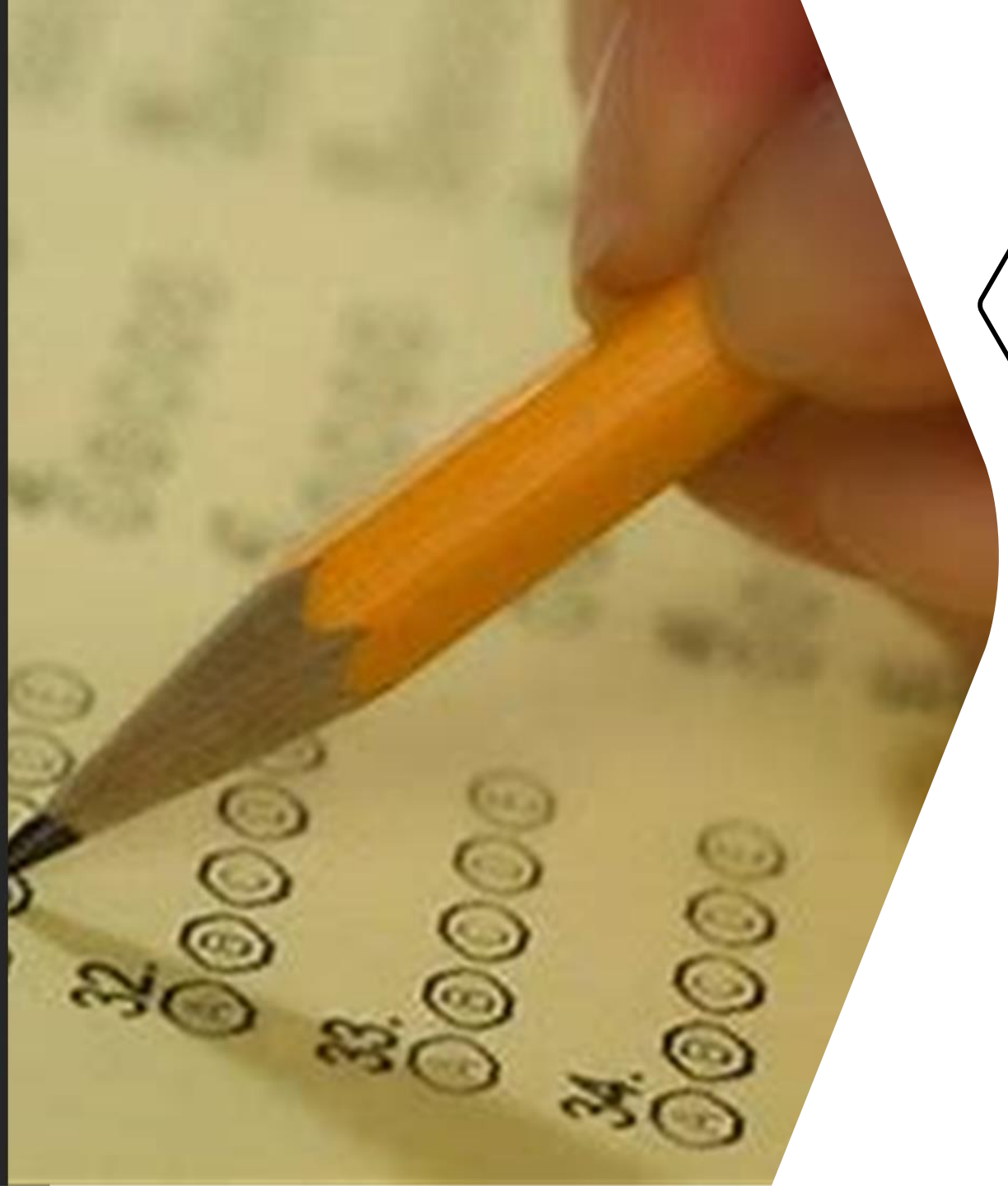


Fourteen-year-old girl , known case of type 1 diabetes for the last 4 years, started to complain from repeated attacks of hypoglycemia, malaise, fatigue, and weight loss. Which one of the following is the most important investigation you are going to order?

a) Measurement of acute phase reactants (ESR & CRP).
b) Measurement of cortisol & adrenocorticotropic hormone.
c) Thyroid autoantibodies.
d) Measurement of C- peptide for possible honey- moon phase.

Primary Adrenal insufficiency

- Is suspected on the basis of clinical features including: fatigue, nausea, vomiting, hypotension, dehydration, electrolyte abnormalities, and may present with adrenal crisis.
- Recurrent unexplained hypoglycemia is indication to screen for adrenal insufficiency.
- Unexplained anorexia with weight loss is another important complaint.
- Laboratory testing reveals low cortisol secretion when tested in the morning (usually at 8 am).
- ACTH is high in the setting of low cortisol, with hyponatremia, hyperkalemia, hypoglycemia, metabolic acidosis and elevated plasma renin activity.

Newborn with genital ambiguity, noted to have generalized hyperpigmentation. Pelvic ultrasound demonstrated the presence of a both ovaries &uterus. What is the most useful investigation?

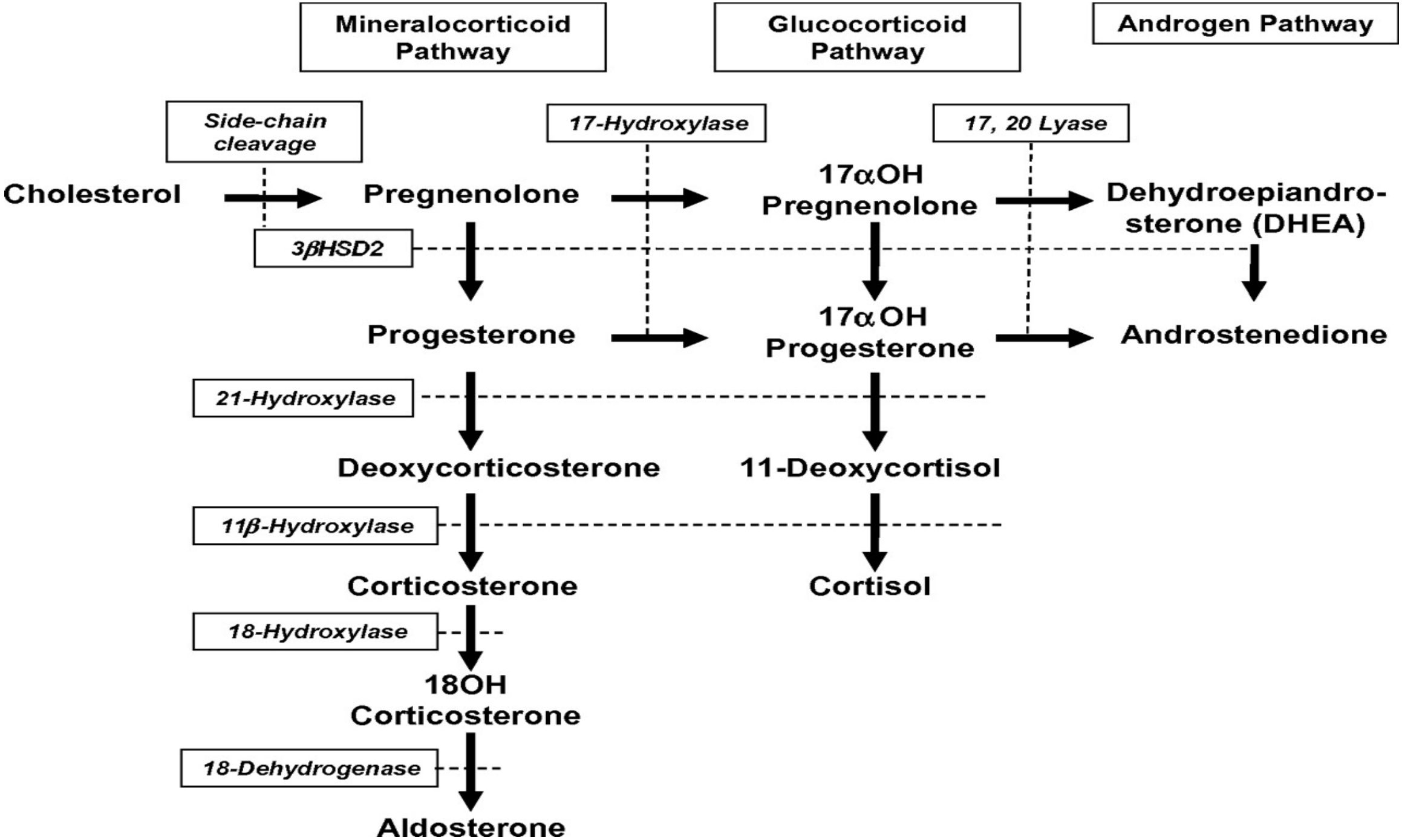
a) 21-hydroxylase enzyme.
b) 17-hydroxyprogesterone.
c) Serum electrolytes.
d) DHEAS.

Congenital adrenal hyperplasia

- It is autosomal recessive disorders.
- Over 95 % of cases are due to 21-hydroxylase deficiency.
- We can’t measure the deficient enzyme, but we measure the biochemical consequences of deficient enzyme.
- In many countries, neonatal screening for 21OHD is performed routinely in all newborns by measurement of 17OHP in a dried, filter paper blood spot which will be elevated.
- False-positive results are common with premature infants (reference ranges based on weight & gestational age) should be avaliable.
- False-negative results may occur as a result of:
  - antenatal maternal glucocorticoid use,
  - in first 2 days of life.
  - in sick neonates.

Ten-day old boy, presented with hypertension & metabolic alkalosis. He was diagnosed with congenital adrenal hyperplasia. Which of the following enzyme deficiency could be the cause?

a) Aldosterone synthase deficiency.
b) 21-hydroxylase deficiency.
c) 3-β-hydroxysteroid dehydrogenase deficiency.
d) 17- hydroxylase deficiency.


Mineralocorticoid Pathway
Glucocorticoid Pathway
Androgen Pathway
Side-chain cleavage
17-Hydroxylase
17, 20 Lyase
Cholesterol
Pregnenolone
17αOH Pregnenolone
Dehydroepiandro-sterone (DHEA)
3βHSD2
Progesterone
17αOH Progesterone
Androstenedione
21-Hydroxylase
Deoxycorticosterone
11-Deoxycortisol
11β-Hydroxylase
Corticosterone
Cortisol
18-Hydroxylase
18OH Corticosterone
18-Dehydrogenase
Aldosterone

- 17-Hydroxylase (17-OH) deficiency is rare form of congenital adrenal hyperplasia.
- It causes decreased production of glucocorticoids & sex steroids, resulting in 46,XY DSD.
- Increased synthesis of mineralocorticoids precursors, resulting in hypertension & hypokalaemia
- Exogenous glucocorticoid therapy is the treatment of choice which suppresses (ACTH) secretion, decreases 11-DOC & corticosterone levels, and normalizes serum potassium and blood pressure.

Six-month old 46 XY infant presented with non palpable testes.
Which one of the following, the most appropriate next step would be?

a) Assure & re-examination him again at age 18 months.
b) Refer the patient for exploratory laparotomy.
c) Measure basal & HCG stimulated testosterone & DHT.
d) measure serum level of Anti-Müllerian hormone (inhibin B).

Cryptorchidism

- Usually is an isolated finding.
- It may be secondary to endocrine disorders, genetic syndromes, &embryological abnormalities, particularly if the cryptorchidism is bilateral.
- Bilaterally nonpalpable testes in a phenotypically male newborn require immediate evaluation at the time of birth because the differential diagnosis includes DSD.
- Anti-Müllerian hormone (inhibin B), is well-accepted sertoli cell biomarker to evaluate testicular function during childhood without the need for stimulation tests.
- Prepubertal boys with cryptorchidism, especially those with bilaterally undescended gonads, have decreased AMH production.
- Although serum AMH may fall within the normal range, there is a considerable prevalence of testicular dysfunction during childhood in this frequent condition.

Neonate was born with abnormal newborn thyroid screen done at 3 days of age: fT4 was 8 pmol/l (12-22), TSH was 60 µIU/mL (0.3-5.0). Which one of the following might be the cause?

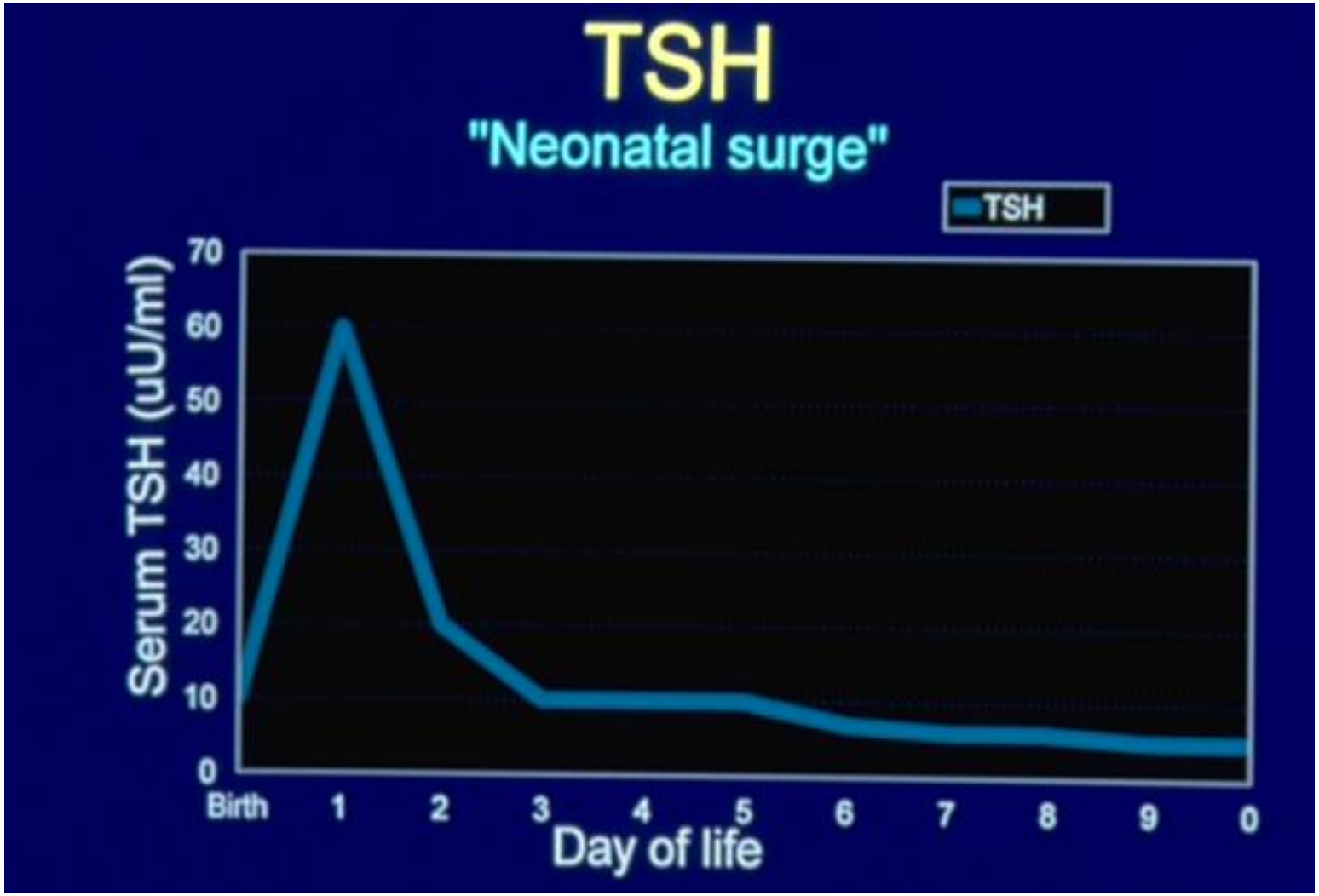
a) Hypothyroidism due to dysgenesis of the thyroid gland.
b) Central hypothyroidism.
c) TBG deficiency.
d) Hypothyroidism from excess iodine exposure.

Congenital Hypothyroidism

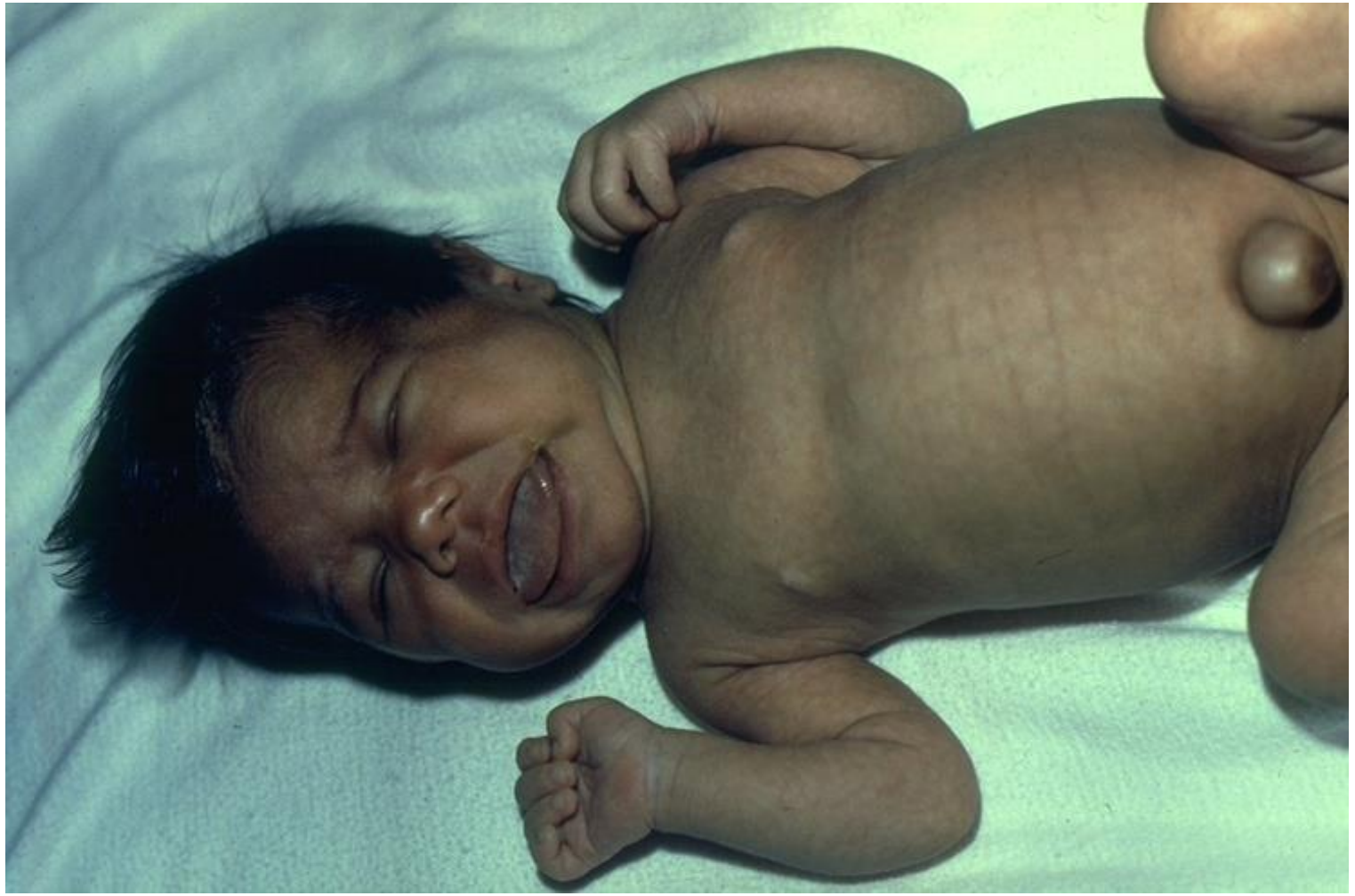
- Thyroid dysgenesis/agenesis is the commonest cause of congenital primary hypothyroidism in almost 90% of cases.
- Prevalence 1 in 4,000.

Clinical manifestations:

- Decreased activity.
- Large anterior fontanelle.
- Poor feeding.
- Poor growth.
- Prolonged neonatal Jaundice or yellow skin.
- Constipation.
- Hypotonia.
- Hoarse cry.


TSH
"Neonatal surge"
TSH
Serum TSH (uU/ml)
70
60
50
40
30
20
10
0
Birth
1
2
3
4
5
6
7
8
9
0
Day of life

Congenital Hypothyroidism



A preterm baby, 28 weeker with birthweight of 1,2 kg. Thyroid screening test revealed low fT4 and low TSH. Which one of the following is the most accurate statement?

a) Hypothalamic-pituitary-thyroid axis immaturity.
b) Hypothyroxinemia of prematurity needed short term Thyroxine replacement.
c) Sick euthyroid syndrome.
d) Abnormal thyroid function due to application of iodine-containing antiseptics.

Hypothyroxinemia of prematurity

- Preterm infants with low T4 & low TSH concentrations on the initial screening may have physiologic immaturity of the hypothalamic-pituitary-thyroid axis.
- In the majority of extreme prematures, T4 will rise into normal range when a second (or third) screening test is performed at 2-4 weeks of age, as hypothalamic-pituitary-thyroid function matures.
- Recommendation for three screening test in very preterm infants (e.g., birth weight <1500 g or gestational age <32 weeks).
  - In many programs, three screening tests are performed (at 24 to 48 hours, 10 to 14 days, and 4 to 6 weeks of age).
- Thyroxine treatment is matter of debate whether or not is beneficial.

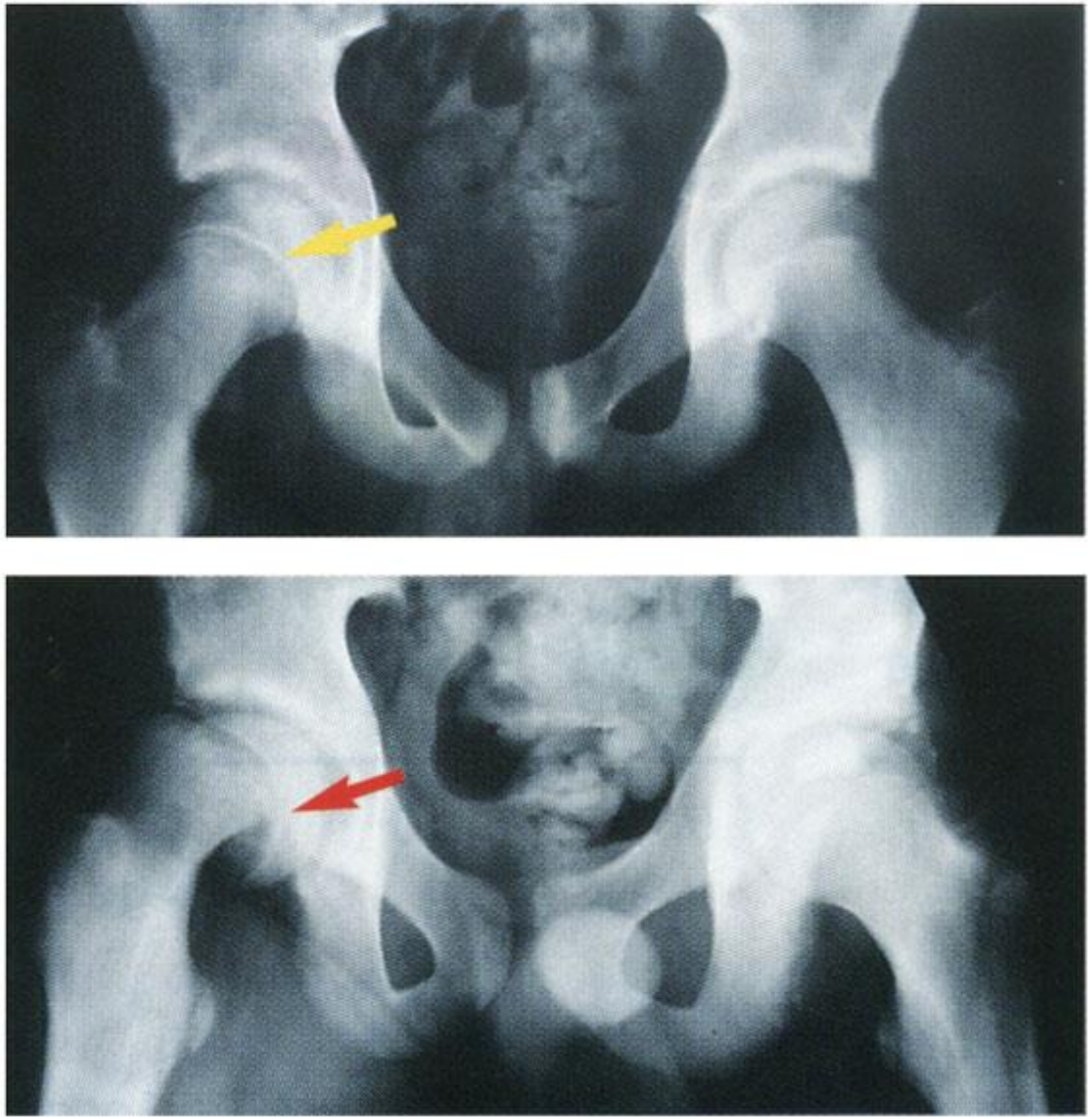
Thirteen -year-old - girl presented to the orthopedic clinic with two year history of a limping in her left leg. She has been diagnosed with hypothyroidism but not compliant on her medication. On examination she is short & obese with a goiter and other signs of hypothyroidism. She had limitation of movement of her left hip and she is limping. What is the most likely diagnosis?

a) Slipped capital femoral epiphysis (SCFE).
b) Chronic Osteomyelitis.
c) Vitamin D deficiency.
d) Monoarticular rheumatoid arthritis.

Slipped capital femoral epiphysis (SCFE)

- Characterized by displacement of the capital femoral epiphysis from the femoral neck through the physeal plate.
- The mean age of presentation is 12 years in girls and 13 years in boys, near the time of peak linear growth.
- Obesity is a significant risk factor.
- The two most common features of the presentation are pain and altered gait.
- Usually chronic or intermittent, non radiating, dull, aching pain in the hip, groin, thigh, or knee, and no history of preceding trauma.
- The diagnosis is usually made on plain radiographs, which reveal an apparent posterior displacement of the femoral epiphysis, like ice cream slipping off a cone.

Slipped capital femoral epiphysis (SCFE)



Eight –year - old girl, has presented with goiter, together with high TSH, low fT4. She has sensorineural hearing loss. What is the most likely inheritance of her disease?

a) Autosomal recessive.
b) Autosomal dominant.
c) Sex-linked dominant.
d) Sex –linked recessive.

Pendred syndrome

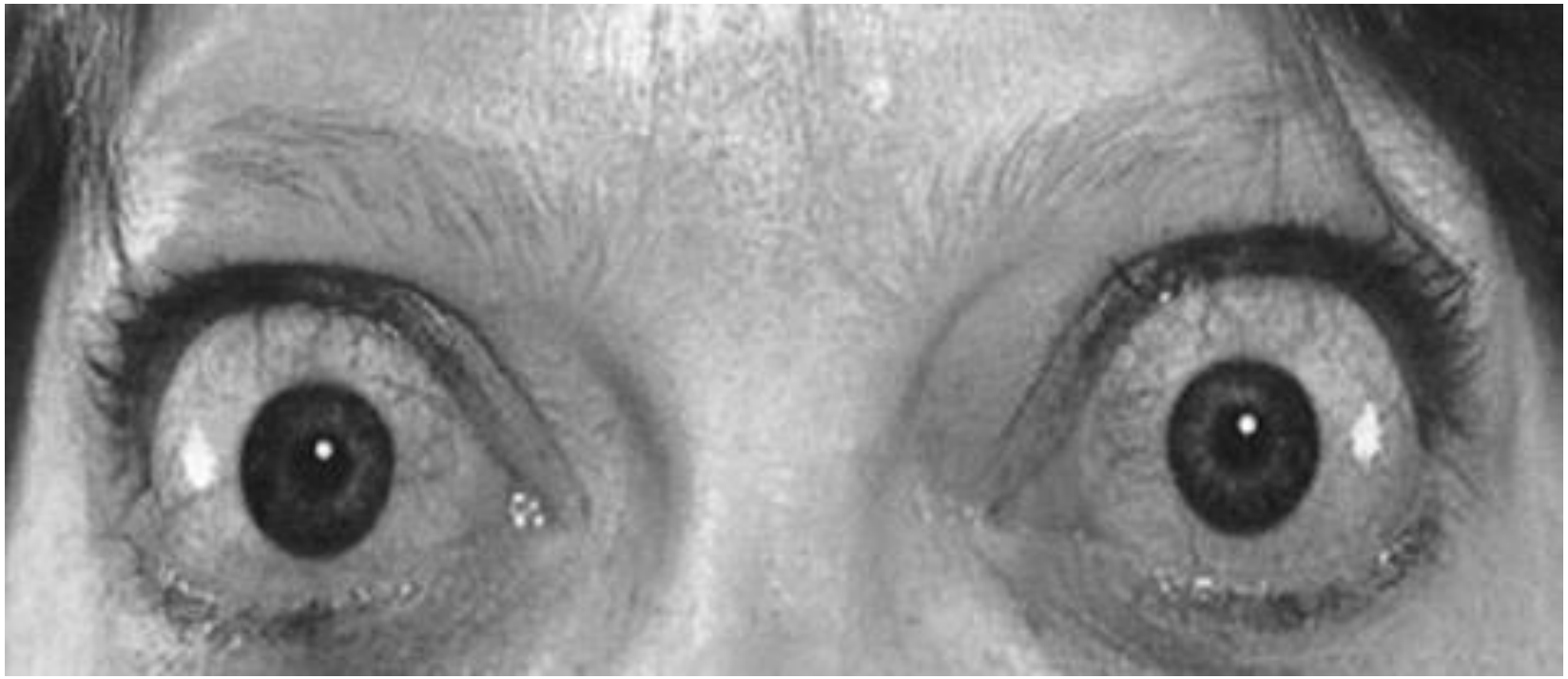
- It was named after “British” Dr Vaughan Pendred (1869–1946).
- Autosomal recessive.
- Is caused by a mutation in the *SLC26A4* gene, which causes a defect in the organification of iodine (i.e., incorporation into thyroid hormone), congenital sensorineural hearing loss, and goiter.
- Combination of congenital bilateral sensorineural hearing loss & goiter with euthyroid or mild hypothyroidism.

Twelve-year old girl, who has presented with exophthalmos & weight loss. Her laboratory tests confirm Graves disease. Which one of the following is the most likely cause for her exophthalmos?

a) Fat tissue deposition.
b) Ocular muscle hyperplasia.
c) Inflammatory infiltrate.
d) Globe enlargement.

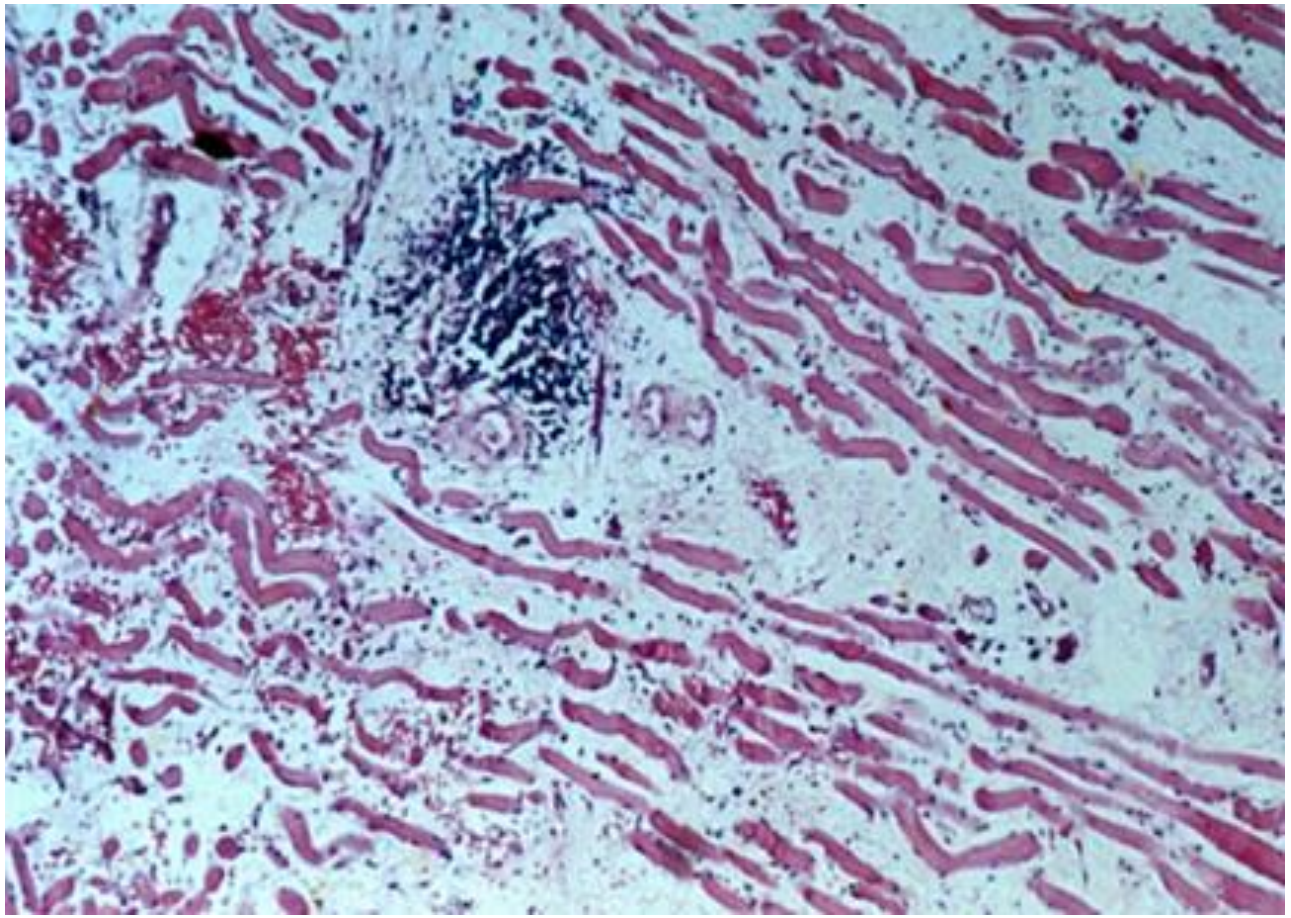
Eye signs of Grave's disease

- Staring look.
- A gritty or foreign object sensation in the eyes.
- Excessive tearing.
- Lid lag.
- Lid retraction.
- Chemosis.
- Ophthalmopathy.
- Eye or retroocular discomfort or pain.
- Blurring of vision.
- Diplopia.
- Occasionally decreased vision.



Graves' orbitopathy (ophthalmopathy)

- Is an autoimmune disease of the retroorbital tissues.
- There is no much evidence that RAI causes or worsens Graves ophthalmopathy in children or adolescents, as compared to adults.
- Children with mild orbitopathy may still be candidates for anti-thyroid, radioiodine, or surgery.
- Children with moderate-to-severe orbitopathy, anti-thyroid or surgery are the preferred treatment options.
- Patients who refuse surgery and who have contraindications to anti-thyroid may need to be offered radioiodine therapy with steroid coverage.
- Prednisone (1 mg/kg/day) for 4 weeks, followed by gradual tapering over another 2-3 months depending on severity and response.



Muscle fibers show loss of striation, fragmentation, and infiltration with lymphocytes, most of which are T lymphocytes.

Thirteen-year-old girl presented with a history of loss of weight, dizziness and palpitations over the past two weeks. On examination, she was warm, pulse rate 115 bpm. Heart auscultation with audible bruit. Thyroid function test revealed picture of hyperthyroidism. Which one of the following investigation is useful in confirming the diagnosis of Graves disease?

a) Thyroid stimulating antibodies.
b) Ultrasound scan of the neck.
c) Anti-thyroglobulin antibody.
d) Antithyroid peroxisomal antibody.

Graves disease

- Clinical manifestations of tachycardia, weight loss or poor weight gain, and lid lag, tremor and neuropsychiatric symptoms.
- Most patients have goiter & ophthalmopathy.
- The diagnosis of thyrotoxicosis is confirmed by the findings of elevated fT4 and fT3, with suppressed TSH.
- The diagnosis of Graves disease as the cause of the thyrotoxicosis is confirmed by the presence of thyrotropin receptor antibodies (TSHR-Ab), which are detectable in the majority of children with Graves disease.

Ten – year old girl with graves disease, on carbimazole therapy.
Which one of the following is the most serious side effect of anti thyroid medication?

a) Fever.
b) Rash.
c) Granulocytopenia.
d) Arthritis.

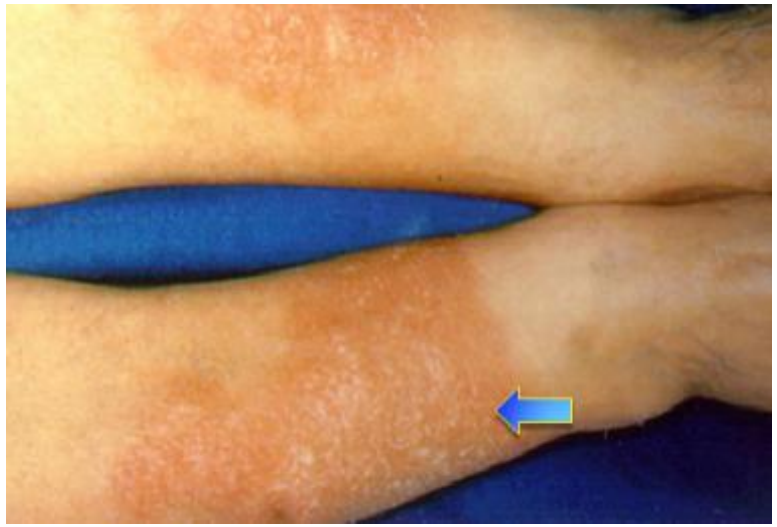
Anti-thyroid therapy

- Thionamide compounds were found in 1943 to inhibit thyroid hormone synthesis.
- Methimazole (MMI) is first choice of anti- thyroid therapy.
- PTU is associated with risk of severe hepatotoxicity.
- Anti –thyroid medications, inhibit both organification of iodine to tyrosine residues in thyroglobulin & coupling of iodothyronines.
- Adverse effects of anti- thyroid therapy include:
  - minor side effects (e.g., rash, fever, arthritis)
  - rare serious adverse effects, such as agranulocytosis & hepatotoxicity.
- If remission does not occur following ATD therapy, ^{131}I or surgery should be contemplated.

Eleven- year- old boy confirmed to have Graves disease, has presented with severe proptosis , tachycardia & fine tremor. Which of the following is most accurate statement?

a) Radioactive iodine therapy in this case is first line of management.
b) PTU is more preferable than MTZ.
c) Agranulocytosis is a complication of anti thyroid medications.
d) Surgery is a good option for this child.

Sixteen- year - old boy presented with thyrotoxicosis with bilateral, firm, non-pitting, asymmetrical plaques over his legs (see photo).



Which one of the following describes skin lesion?

a) Pretibial myxedema.
b) Scleroderma.
c) Eczematous lesions.
d) Dermatomyositis.

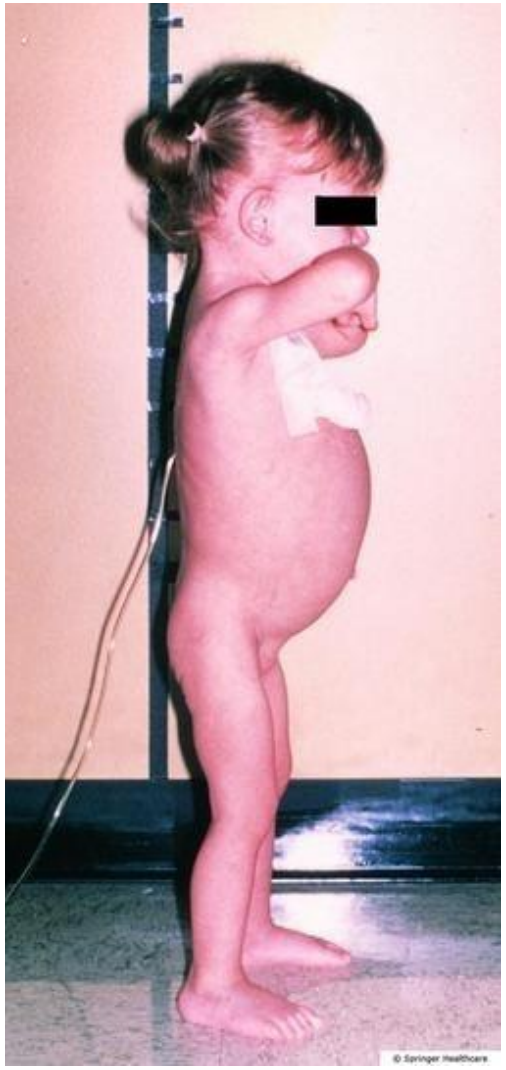
Pretibial myxedema (PTM)

- Thyroid dermopathy occurs rarely.
- PTM is term used to describe localized lesions of the skin resulting from the deposition of hyaluronic acid, usually as a component of thyroid disease.
- Although PTM is most often confined to the pretibial area, it may occur anywhere on the skin, especially the ankle, dorsum of the foot, knees, shoulders, elbows, upper back, pinnae, nose, and neck.
- It is nearly always associated with autoimmune thyroid disease.

Six-year-old girl is referred with growth failure, poor appetite, recurrent abdominal pain, ‘thick custard’ stools and vomiting.
What is the appropriate most confirmatory investigation?

a) Bone age.
b) Anti-tissue transglutaminase antibody.
c) Duodenal /Jejunal biopsy.
d) Serum iron & ferritin.

Remember: if a child presented with abdominal distension & short stature, think of celiac disease !



Celiac disease

- Children with celiac disease may exhibit no unusual findings on physical examination.
- Sometimes, protuberant abdomen & marked muscle wasting and evidence of malnutrition.
- Usually, they responded very well to a strict gluten-free diet that she must follow for the rest of her life.
- In addition, total IgA levels should be measured.
- IgA - anti-tissue transglutaminase (tTG) antibody is the single preferred test for detection of celiac disease.
- Patients with positive serologic testing, should undergo an upper endoscopy with small bowel biopsy to diagnose celiac disease.
- In patients with IgA deficiency, IgG-deamidated gliadin peptide (DGP) IgG.

Good LUCK

End of MCQs Revision
Part 2