Pediatric Endocrinology Review MCQs

PART - 4

Abdulmoein Eid Al-Agha, FRCPCH
Professor of Pediatric Endocrinology,
Website: http://aagha.kau.edu.sa

Seventeen - year old adolescent girl, presented with hirsutism, irregular menses and acne. Which of the following supports the diagnosis of PCOS?

a) High FSH level.
b) LH: FSH ratio of 3:1.
c) LH: FSH ratio of 1:1.
d) High estradiol level.

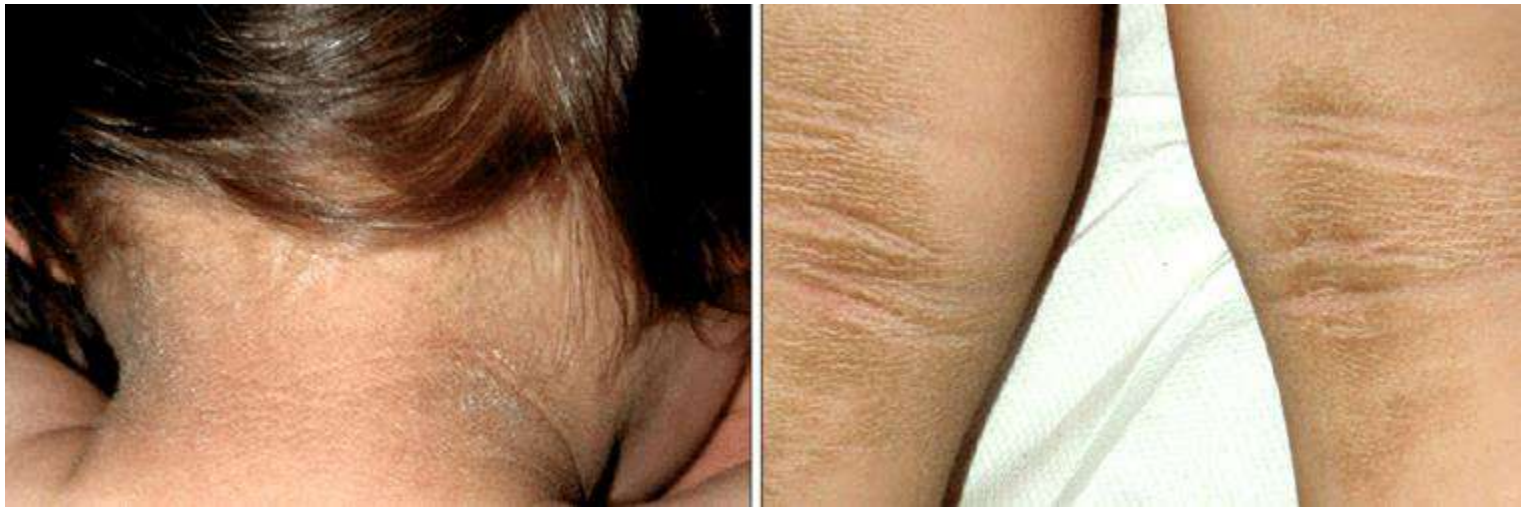
Definitions

- Primary amenorrhea: Lack of menses by 16 years of age.
- Secondary amenorrhea: More than 90 days without a menstrual period, after previously menstruating.
- Irregular menstruation cycle: if menstrual cycle is less than 24 days or more than 38 days.
- Irregular menstruation is common in adolescence especially within a year of first menses.

PCOS

- Most expert groups use “Rotterdam criteria” for the diagnosis of PCOS.
- Two out of three of the following criteria are required to make the diagnosis:
  - Oligo- and/or anovulation.
  - Clinical and/or biochemical signs of hyperandrogenism.
  - Polycystic ovaries (by ultrasound).
- Other causes of hyperandrogenism should be excluded:
  - non-classic congenital adrenal hyperplasia.
  - hyperprolactinemia.
  - Cushing syndrome.
  - androgen-secreting ovarian / adrenal tumors.

Thirteen –year old girl, who was presented with virilization& irregular menses for the last 6 months. Her parents are first cousin. On general examination(photos). Which one of the following, is most likely diagnosis?

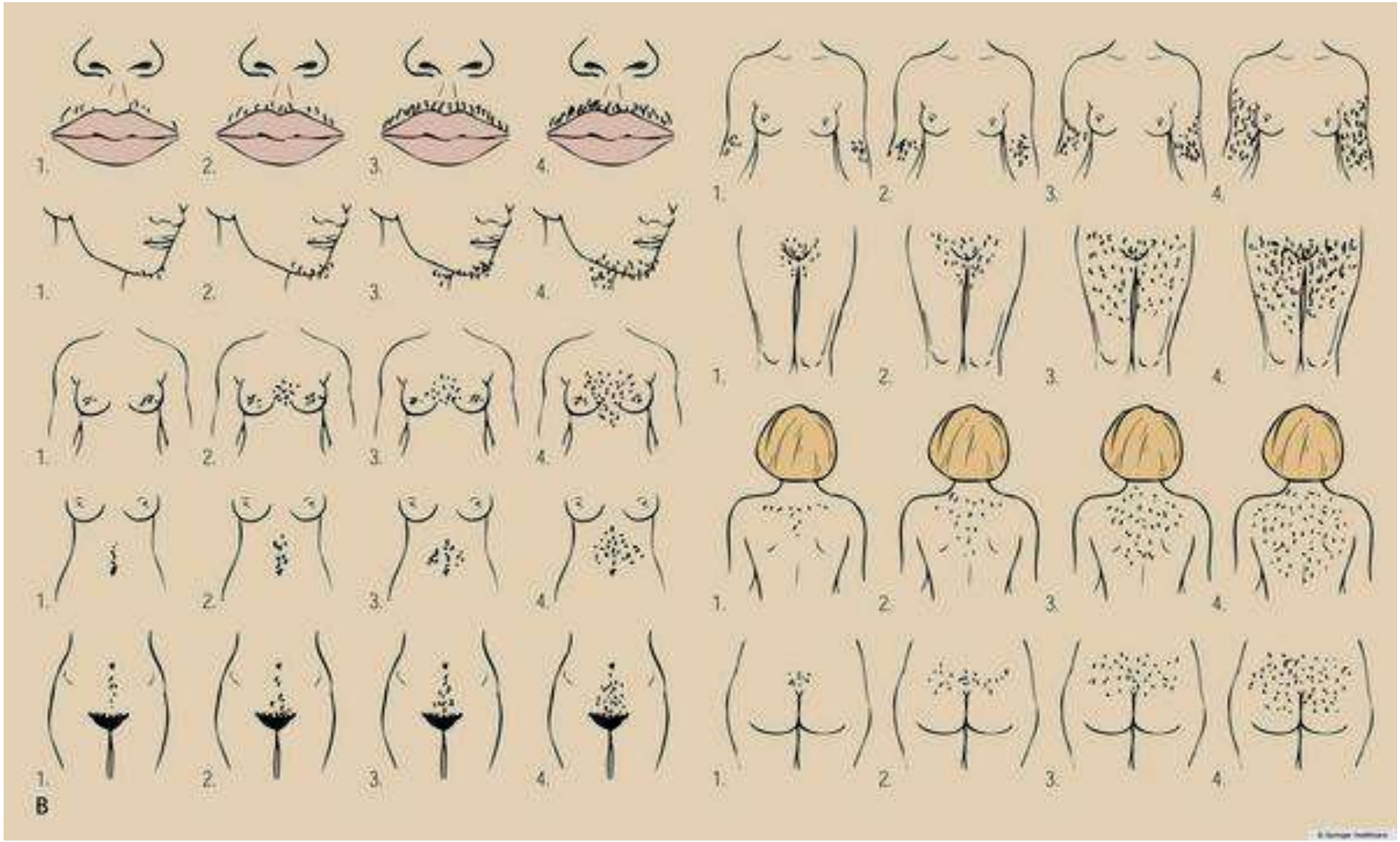


a) Cushing syndrome.
b) Polycystic ovary disease.
c) Virilizing adrenal tumor.
d) Non-classical CAH.

Hirsutism Vs virilization

- Virilization includes clitoromegaly, male-pattern baldness, deepening of the voice, and increased muscle mass in addition to hirsutism and chronic anovulation.
- The magnitude of hirsutism could be scored by Ferriman-Gallwey score.
- score assesses hair growth in nine androgen-dependent areas (grades 1 to 4) from which a score is derived (total 36 scores).
- Grade 1 indicates minimal terminal hair growth and grade 4 indicates dense terminal hair growth.
- Scores ≥ 8 are considered to indicate hirsutism.

Ferriman-Gallwey score



Which one of the following medications is used in treatment of hirsutism?

a) Thiazide diuretic.
b) Oral contraceptives.
c) Sildenafil.
d) Minoxidil.

- Contraceptives would treat hirsutism in 2/3 of cases.
- Direct suppression of ovarian steroid production.
- Increase hepatic binding globulin synthesis, which binds circulating androgens lead to decrease free androgens levels.

Sixteen - year –old phenotypically female, presented with primary amenorrhea. Height 160 cm, weight 45 kg. Tanner stage of B3 PH 1-2. What is the most likely diagnosis?

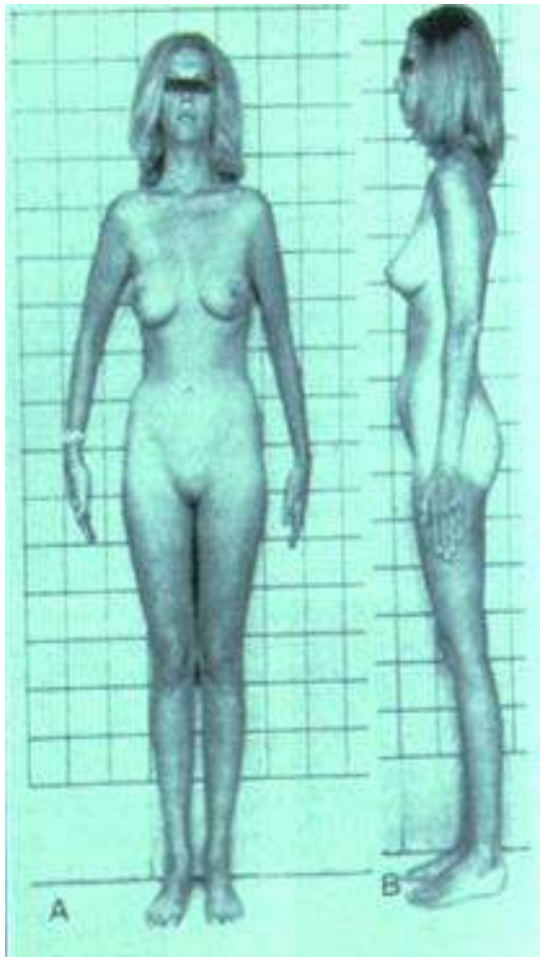
a) Mullerian dysgenesis.
b) Turner’s syndrome.
c) Complete androgen insensitivity syndrome.
d) Kalman's syndrome.

Complete androgen insensitivity syndrome (CAIS)

XY Genotype

Testosterone → ~~*Androgen Receptor*~~

Testosterone —Aromatase→ **Estradiol** → *Estrogen Receptor*



Complete androgen insensitivity syndrome(AIS)

- Formerly known as testicular feminization.
- X-linked recessive condition.
- Resulting in a failure of normal masculinization of the external genitalia in 46 XY.
- Males with complete androgen insensitivity syndrome have completely normal female external genitalia.
- Affected males have normal testes with normal production of testosterone and normal conversion to dihydrotestosterone (DHT).
- No fallopian tubes, uterus, or upper vagina.

Definition of delayed puberty

- In boys:
  - No enlargement of the testes (testicles) by age 14 years.
- In girls:
  - No breast development by age 13 years.
  - A time lapse of more than 5 years from the beginning of breast growth to the first menstrual period.
  - No menstruation (amenorrhea) by age 16 years.

Twelve –year old girl, who has presented with height below 3rd%, with growth velocity of 3 cm/ year. Both parents were of average height (MPH 168 cm between 10th-25th%). On examination, Tanner staging was B3 PH 2. Among which one of the following is most appropriate approach?

a) Observe & follow up in next six months.
b) Re-assure the family, shortly she will catch- up.
c) Do bone age.
d) Do full hormonal assessments workup.

Short stature

- Defined as height 2 standard deviations below the mean height for children of that sex and chronologic age in a given population.
- Is essential to differentiate physiological from pathological causes.
- To differentiate, physiological from pathological short stature, is essential to calculate growth velocity, MPH & bone age assessment.
- Laboratory evaluation should be done in pathological short stature.
- Children with pathological short stature should be evaluated with CBC, ESR, tTG IgA, creatinine, electrolytes, TSH, free thyroxine, IGF-1, and IGFBP-3.
- A karyotype in any short girl, to exclude Turner's syndrome.
- Skeletal survey for children suspected skeletal dysplasia.
- If GH deficiency is suspected a proactive test using 2 pharmacological stimualtion.

Six- year old boy, known case of growth hormone deficiency on GH therapy, referred from orthopedic clinic for second opinion, as he has thoraco-lumbar scoliosis. Which one of the following is the best management choice?

a) No worries, continue on GH safely.
b) Stop GH therapy immediately.
c) Decrease the dose of GH.
d) Continue with close observation, with follow up with orthopedic surgeon.

Scoliosis & GH therapy

- Scoliosis related to rapid growth that occurs with therapy and is not a direct effect of the growth hormone.
- Patients with scoliosis who are treated with growth hormone should have their scoliosis monitored during therapy.
- No clear statement to stop GH therapy, rather close monitoring during therapy.

Contraindications of GH therapy

- GH should not be used, if patient's epiphyses are closed.
- Active proliferative or severe non-proliferative diabetic retinopathy.
- In patients with any evidence of any tumor.
- In patients pre-existing or active malignancy.

Eight-year-boy recovered from medulloblastoma 4 years ago. Treatment at that time consisted of chemotherapy & craniospinal irradiation. Child' clothes of same size for the last one year. On examination, height was below 3rd%, weight on 25th %. His upper: lower segment was 0.8:1. Which one of the following is the most likely cause of short stature?

a) Acquired growth hormone deficiency.
b) Chemotherapy-induced hypopituitarism.
c) Irradiation-induced spinal epiphyseal fusion.
d) Acquired central hypothyroidism.

Craniospinal irradiation

- Is part of treatment of medulloblastoma.
- Has profound effect on spinal growth.
- The younger the child is when given irradiation the greater the subsequent skeletal disproportion.
- Radiation induced growth hormone (GH) deficiency and spinal irradiation are two major adverse factors that contribute to the short adult height achieved by many patients treated for brain tumors in childhood.

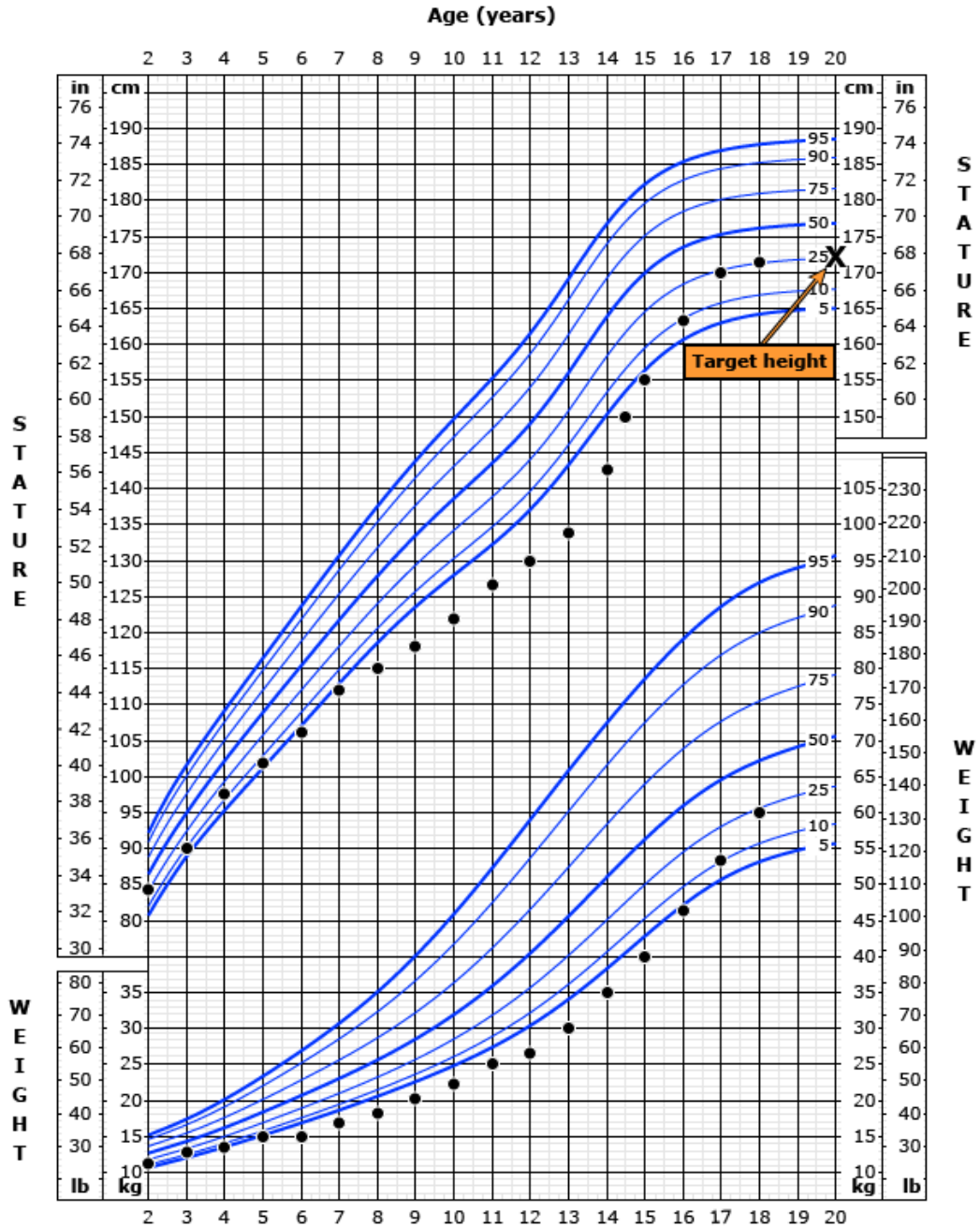
Fourteen - year-old boy, who was growing well till 2 years ago, when his family noticed slow growth. He is an otherwise healthy child. On his examination, his height percentile dropped below 3rd. percentile in comparison to 25 %, 2 years ago. His Tanner's stage was prepubertal. Bone age is greater than two standard deviations below the chronologic age. Which one of the following, is the MOST likely cause of short stature?

a) Familial short stature.
b) Growth hormone deficiency.
c) Constitutional delay.
d) Hypogonadism.

Constitutional delay of growth and puberty (CDGP)

- Children with CDGP usually are growing at a low-normal rate (e.g., about 4 to 5 cm/year in preadolescent girls, and 3.5 to 4.5 cm/year in preadolescent boys).
- In addition to a low preadolescent height velocity, they tend to have delayed pubertal development.
- This leads to a marked height discrepancy during the early teenage years compared with their peers, but is followed by catch-up growth when they do enter puberty with normal final adult height.
- In many cases, there is a family history of delayed growth and puberty in one or both parents (sometimes described as being a "late bloomer").

Constitutional delay of growth and puberty



Which one of the following disorders of growth is characterized by normal body proportion ?

a) Achondroplasia.
b) Morquio’s syndrome.
c) untreated congenital hypothyroidism.
d) Hypopituitarism.

Which one of the following is not a proven adverse effect of GH replacement therapy?

a) Carpal tunnel syndrome.
b) Arthralgia and myalgia.
c) Benign intracranial hypertension.
d) Increase incidence of brain tumor.

Increase incidence of brain tumor is never documented & is not proven adverse effect of growth hormone therapy.

Three years old boy presented with goiter, short stature, deafness and symptoms suggestive of mild hypothyroidism. On examination, is having normal mentality, diffuse goiter, deaf and mute with normal CNS examination apart from sluggish reflexes. He has high TSH, as well high fT4 and fT3. Which of the following is most likely diagnosis?

a) Generalized resistance to thyroid hormone (GRTH)
b) Pituitary resistance to thyroid hormone (PRTH)
c) Pendred's syndrome
d) TSH secreting Adenoma

Resistance to thyroid hormone (RTH)

- Is usually dominantly inherited.
- Characterized by elevated fT3 &fT4 and failure to suppress TSH secretion.
- Variable refractoriness to hormone action in peripheral tissues.
- Two major forms:
  - asymptomatic individuals with generalized resistance (GRTH).
  - patients with thyrotoxicosis features, suggesting predominant pituitary resistance (PRTH).
- The characteristic blood test results for this disorder can also be found in other disorders (for example TSH-oma (pituitary adenoma), or other pituitary disorders).
- The diagnosis may involve identifying mutation of the thyroid receptor, which is present in approximately 85% of cases.

Seven-year-old girl, referred because of tall stature. On examination, no dysmorphic features. Height was > 97% , MPH was between 50- 75%. Tanner staging B2, PH3. Bone age of 12 years. What is your initial approach?

a) Most likely familial need to observe growth velocity.
b) Need to do basal & stimulated GH test.
c) Need to do LH, FSH & estradiol levels.
d) Need to do oral glucose tolerance test for GH suppression.

Tall stature

- Height > 2 SD above the mean height for age, sex and race.
- Obesity leads to tall stature in childhood.
- Other causes of tall stature, include excessive secretion of growth hormone or sex hormones because of precocious puberty.
- In precocious puberty, initially, will be early growth acceleration, leading to tall stature, followed by early epiphyseal closure, resulting in short stature as adulthood.

Four- month old infant, presented with failure to thrive. His mother was complaining of too many diaper change & urine was leaking out of diapers most of the time. On examination he was having, moderate to severe dehydration. His initial sodium was 175 mmol/l, urine osmolality 105 mosmol/l, serum osmolality 315 mosmol/l. Which one of the following is most common cause in the differential diagnosis of this infant?

a) Langerhans cell histiocytosis.

b) X-linked dominant nephrogenic DI.

c) DIDMOAD syndrome.

d) Psychological polydipsia.

Congenital nephrogenic diabetes insipidus

- Two genetic mutations have been identified causing nephrogenic diabetes insipidus present at birth.
  - Vasopressin receptor gene mutation (AVPR2) is responsible for 90% of all cases of congenital diabetes insipidus, sex linked dominant (happens in boys).
  - The remaining 10% of cases, caused by AQP2 gene mutation (responsible for water reabsorption in response to ADH, autosomal recessive (affect both boys & girls)

GOOD LUCK

End of part- 4