Thyroid physiology and congenital hypothyroidism – Q&A

Question #1

Of the following choices, which is the most frequent cause of congenital hypothyroidism?

A) Organification defect
B) Sodium-iodine symporter defect
C) Sublingual thyroid gland
D) TSH receptor defect

Answer: C

Thyroid dysgenesis (including a sublingual gland) is the most common cause of congenital hypothyroidism.

Question #2

You are consulted on a 9-day-old premature female infant with cleft palate for abnormal thyroid function tests. Her initial newborn screen was normal. Due to problems with low blood sugars, various labs were performed including thyroid studies: TSH = 3.2 mU/mL and free T4 = 0.3 ng/dL. Which of the following hormone deficiencies is this infant most likely at risk of having at this time?

A) ACTH
B) ADH
C) GH
D) LH

Answer: C

The infant's labs are concerning for central hypothyroidism. In the setting of hypoglycemia and cleft palate, one should be most concerned about growth hormone deficiency.

Question 3

You are called about an 8-year-old male with known congenital hypothyroidism. He is being seen in a local Emergency Department due to the recent onset of diarrhea, irritability, spasms and contractures of his hands and feet. You are also told that an EKG revealed a prolonged QT interval. Review of his past records show that his TSH at diagnosis of congenital hypothyroidism was 18 mU/mL, and he has been well controlled on 44 mcg of levothyroxine daily. The most likely reason to explain this patient's congenital hypothyroidism is due to an inactivating mutation in which of the following genes?

A) GNAS
B) NIS
C) PDS
D) TSHR

Answer: A

The patient has a history of congenital hypothyroidism and is presenting with hypocalcemia. This patient most likely has pseudohypoparathyroidism due to an inactivating mutation in GNAS.

Question 4

An 11-year-old male presents to your practice to establish care for management of congenital hypothyroidism. Review of the medical records from his previous provider shows that he had an abnormal newborn screen with an elevated TSH. Serum thyroid function tests confirmed the diagnosis as his TSH = 229 mU/mL at 10 days of life. A sodium pertechtenate 99m (Tc99m) scan at diagnosis was consistent with thyroid agenesis. Mother noted that he stopped taking the levothyroxine about 6 months ago as they were in the process of moving and finding a new doctor. Family history is remarkable for his mother, father, and aunt having hypothyroidism. On exam, his thyroid is not enlarged. Thyroid labs are ordered and his TSH = 2.21 mU/mL and T4 = 9.2 mcg/dL. You then perform a thyroid ultrasound which shows a normal thyroid gland in the normal location. Which of the following is the most likely explanation of the patient's congenital hypothyroidism?

A) Inactivating mutation of the TSH receptor
B) Organification defect
C) Sodium-iodine symporter defect
D) TSH receptor blocking antibodies

Answer: D

All of the answers can result in absent radionuclide uptake, but the only answer that will result in a transient form of congenital hypothyroidism is maternal TRAbs.

Question 5

You are called by a local pediatrician about a 3-week-old infant who has struggled to gain weight. Newborn screen was normal. As part of the evaluation, the pediatrician performed thyroid studies which included a TSH = 6.9 mU/mL and free T4 = 1.7 ng/dL. The best recommendation to give when talking to the pediatrician includes:

A) Check a thyroid ultrasound
B) Recheck thyroid studies in 8 weeks

C) Reassure that the labs are normal
D) Start levothyroxine

Answer: C

The TSH may be flagged as being elevated for the lab's reference range, but in a 3 week old infant, the thyroid labs are normal for age.