1. The parents of a 10-year old girl are concerned about her height. She is the shortest in her class and they request growth hormone therapy. Which of the following is true regarding the diagnosis/management of her condition?

- a. A skeletal survey is needed to rule out hypochondroplasia
- b. Growth hormone therapy is indicated because she may have growth hormone deficiency
- c. The parents should be reassured that she will have catch-up growth during the adolescent growth spurt
- d. Chromosomal analysis for karyotyping will reveal the diagnosis
- e. She has constitutional growth delay

**Turner Syndrome**

- 45XO/ 15% due to mosaicism/ major locus involved in the control of linear growth/3% of all pregnancies
- Lymphedema/loose skin folds/webbing of the neck/ broad chest/short stature/ early menopause
- Cardiac defects: coarctation (20%) more so in the web neck
- Renal: horseshoe kidneys, pelvic kidney, UPJ obstruction/ Idiopathic hypertension
- Recurrent otitis (75%) mild neurological problems
- Endocrine- thyroid/bone abnormalities:
- FSH/ultrasound of kidneys and heart/ Thyroid studies/ hearing tests/ normal growth hormone levels
- Treatment: recombinant growth hormone/ Estrogens
• Hypochondroplasia: milder/apparent at childhood with stocky build/frontal bossing
• Genetic short stature versus Constitutional growth delay: parental heights/bone age assessment
• Primary hypothyroidism
• Systemic conditions: IBD
• Emotional deprivation: mimics hypopituitarism
• Silver-Russell syndrome: Frontal bossing, triangular facies, hemihypertrophy, shortened and incurved 5th fingers

2. A 2 week-old male infant is seen in the emergency department with one-day history of vomiting. On examination the infant is lethargic and has mottled skin. The infant’s laboratory data includes Na 121 mmol/L, K 6.1 mmol/L, BUN 40 mg/dL, HCO3 12 mmol/L, and Blood glucose of 40 mg/dL. Appropriate management would include all of the following EXCEPT:

- a. Administering intravenous hydrocortisone
- b. Administering 20ml/kg of 0.9% saline intravenously
- c. Administering dopamine infusion
- d. Sending blood for 17OH-progesterone levels
- e. Administering broad spectrum antibiotics

Congenital Adrenal Hyperplasia
• AR/Deficiency of 21hydroxylase(90%)/Salt wasting and virilizing forms
• Newborn screen is available
• Deficiency of 11beta hydroxylase(6-8%): non salt losing/hypertension is a feature
• female pseudohermaphroditism/precocious puberty/short stature
3. An 8 year-old girl who has insulin dependent diabetes, presents with 3-days history of abdominal pain and vomiting. She was seen in the emergency department one week ago for hypoglycemia and was treated for it. On examination she has rapid respirations, dry mucous membranes and poor peripheral perfusion. Appropriate management would include all of the following, EXCEPT:

- a. Initiating fluid resuscitation judiciously keeping in mind not to exceed the total fluid limit of 4 l/m²/day
- b. Monitoring potassium levels frequently
- c. Sodium bicarbonate therapy may cause intracellular acidosis and not indicated
- d. Administering 0.1u/kg of regular insulin subcutaneously
- e. Advising her to rotate the sites of insulin injections at the time of discharge

DKA

- Fluid and Electrolyte therapy: Slow rehydration and replacement of potassium/ benefit of phosphate replacement is not clear/ no place for alkali therapy
- Insulin therapy: Continuous low dose intravenous infusion especially if there is poor perfusion
- Insulin lipoatrophy: occurs after 6months to 2 years of therapy/ erratic absorption of insulin/incidence is less with more purified insulin/ rotate the injection sites

4. A 2 year-old female toddler is brought into your clinic with a concern regarding the development of pubic hair. Appropriate management should include:

- a. Obtaining a bone age measurement
- b. Reassuring her mother that this is probably benign but needs to be followed up closely
- c. Measuring blood levels of androgens and 17OH-progesterone
- d. All of the above
- e. None of the above
Premature adrenarche

- Appearance of sexual hair before the age of 8 years in girls and 9 years in boys without other signs of maturation
- More frequent in girls
- Early maturational event of adrenal androgen production
- Benign condition/no therapy is necessary
- Atypical cases need full work up including ACTH stimulation test
- Patients are high risk for polycystic ovary disease

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5. A 21-month-old toddler presents for a routine physical examination. You note that there has been excessive weight gain recently. Examination shows a markedly obese child with a blood pressure of 130/90 mmHg. Urine analysis shows glycosuria. Which of the following is the best course of action?

- a. Take a detailed history regarding her food intake and advise on caloric restriction
- b. Order an abdominal ultrasound
- c. Measure serum cortisol in the morning and evening to evaluate the diurnal rhythm
- d. Administer subcutaneous insulin therapy
- e. Measure urinary excretion of free cortisol

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Cushing’s syndrome

- Caused by a functioning adrenocortical tumor. >50% occur in children < 3 years
- Typical clinical features include abnormal masculinization, androgen excess, hypertension, hyperglycemia, and increased susceptibility to infections
- Diurnal variation in cortisol secretion do not develop until 3 years of age
- Urinary free cortisol is almost always increased
- Polycythemia, lymphopenia and eosinopenia
- Dexamethasone suppression test is diagnostic
- Cushingoid syndrome: exogenous administration of corticosteroids
6. A 16 year-old adolescent is losing weight in spite of a good appetite. She is a long distance runner at school and she seems to tire easily. She exhibits emotional lability and her school grades have suffered lately. The next most appropriate plan of action is to:

   • a. Order thyroid function tests
   • b. Refer her for a psychiatric evaluation
   • c. Commence a work up for inflammatory bowel disease
   • d. Hospitalize her for intensive nutritional therapy under supervision
   • e. None of the above

Thyrotoxicosis

   • Peak incidence is during adolescence/ higher in girls (5:1)
   • Earliest signs may be poor school performance and emotional lability
   • Goiter is found in almost all patients
   • Exophthalmos, infrequent blinking, proximal muscle weakness, heart failure and flushed skin with excessive sweating
   • T₃, T₄, and Free T₃ and T₄ are all elevated
   • TSH levels are suppressed/Thyrotropin receptor-stimulating antibody is elevated
   • Measurement of thyroglobulin differentiates hyperthyroxinemia due to exogenous thyroid hormone.

7. A 2 month-old male infant presents to your clinic for routine immunization. The infant exhibits signs of hypothyroidism. The following are true regarding congenital hypothyroidism EXCEPT:

   • a. He may have a history of prolonged physiological jaundice in the neonatal period
   • b. A goiter is almost always present in congenital hypothyroidism
   • c. Plain radiograph of his femur may show absence of distal femoral epiphysis
   • d. The incidence of congenital hypothyroidism is lower in the African-American population
   • e. Thyroid stimulating hormone deficiency is a rare cause of this condition
Congenital hypothyroidism

- Thyroid dysgenesis (90%), Thyrotropin deficiency, defective synthesis of thyroxin, defect of iodine transport
- Incidence is lower in African Americans (1/20000 versus 1/4000)
- Prolongation of physiologic icterus caused by delayed maturation of glucuronide conjugation
- Serum T4 levels are low and TSH is elevated
- Distal femoral epiphysis is normally present at birth but is absent in congenital hypothyroidism
- Thyroxin tablets cannot be mixed with soy formula or iron

Hurler syndrome

- Type 1 MPS/autosomal recessive
- Appears normal at infancy
- Coarse facial features, corneal clouding, skeletal abnormality, hepatosplenomegaly, and short stature
- Developmental delay, hearing loss (both conductive and sensorineural), recurrent URI and ear infections
- Cardiomyopathy, multiple valvular lesions, obstructive airway disease
- Enzyme replacement therapy with recombinant alpha-L-iduronidase is available
A one week-old neonate is recovering from an omphalocele repair. He was a full-term infant born by Caesarian-section with a birth weight of 11lb. He develops a generalized seizure, and his rapid blood glucose measurement is 24mg/dL. Which of the following is true regarding this infant’s condition?

- a. He has increased risk of developing Wilms tumor
- b. He has transient hypoglycemia of the newborn
- c. Measurement of urine ketones will be positive
- d. He has panhypopituitarism
- e. He has hypoadrenalism

Beckwith-Wiedemann syndrome

- Overgrowth malformation syndrome, mutations in the chromosome 11p, occurs in 1 in 13,000 births, hyperinsulinemia is main feature
- Macroglossia, hepatospleenomegaly, nephromegaly, and hypoglycemia secondary to pancreatic beta cell hyperplasia
- Hypoglycemia is usually associated with absence of ketones as in Beta-oxidation defects
- Predisposed to specific subset of childhood neoplasms including Wilms’ tumor, hepatoblastoma and adrenocortical carcinoma

A 3 year-old toddler is seen for bow legs. His blood work reveals: serum calcium of 8.5mg/dl and Alkaline phosphatase 850u/dl. All of the following are true statements regarding this toddler and his diagnosis, EXCEPT:

- a. Anticonvulsant therapy is a recognized cause
- b. Cystic fibrosis needs to be ruled out
- c. Inadequate exposure to the sun may be the etiology
- d. It is rare in low birth weight infants and adolescents
- e. Chronic renal failure is a recognized cause
Ricketts

- Nutritional: inadequate exposure to sunlight or inadequate intake. Unsupplemented dark-skinned infants or the breast-fed infants of mothers unexposed to sunlight
- Likely to develop during rapid growth: LBW infants and adolescents
- Malabsorptive disorders: Celiac disease, cystic fibrosis,
- Anticonvulsant therapy: phenytoin and phenobarbital

11. A 6 year-old male was brought to you with a parental concern of nocturnal enuresis. He has developmental delay and has been gaining weight progressively. On further questioning the parents reveal that he was a small for gestational infant at birth and had feeding problems in first few months of life. All of the following are true regarding the boy’s condition EXCEPT:

- a. He may continue to gain weight and remain obese
- b. He has an increased risk of right heart failure
- c. He may have mild to moderate psychomotor retardation
- d. New onset IDDM may be the cause of nocturnal enuresis
- e. He is at risk to develop precocious puberty

Prader-Willi syndrome

- Sporadic condition/Genetic imprinting/>50% deletion of long arm of chromosome 15. The deleted chromosome is paternally derived in all cases. Maternal disomy.
- Neonatal hypotonia and FTT
- Children between one and six years of age commonly manifest symptoms of hyperphagia with progressive development of obesity
- Complications of obesity (e.g., sleep apnea, cor pulmonale, diabetes mellitus, and atherosclerosis), hypogonadism (osteoporosis), mild to moderate developmental delay, and behavioral issues are common
Prader-Willi syndrome

- Standard diagnostic panel for PWS begins with karyotype and methylation studies, followed by fluorescence in-situ hybridization (FISH), and then microsatellite probes to detect maternal uniparental disomy (UPD).
- Growth hormone secretion is generally blunted in PWS.
- Growth hormone treatment is now approved for all children with PWS and growth failure.
- Children and adolescents with PWS who have a BMI >95th percentile should be screened for the development of type 2 diabetes by measuring fasting blood glucose and/or performing oral glucose tolerance tests.

12. A 12-year-old boy presents with a 2-week history of left eye swelling. On examination he has thoracic scoliosis, café-au-lait spots, and moderate hypertension. All of the following are true regarding his condition EXCEPT:

- a. He may have mild learning disability
- b. Cutaneous nodules develop characteristically during adolescence.
- c. He may have optic glioma on the left side with minimal visual disturbance.
- d. He needs a work up for pheochromocytoma.
- e. All his daughters will be carriers of his condition.

Neurofibromatosis Type 1

- NF 1 prevalence is 1: 4000.
- Six or more café-au-lait spots > 5mm pre puberty and >15mm post puberty (100%).
- Axillary or inguinal freckling.
- 2 or more Lisch nodules.
- One plexiform neurofibroma or 2 or more neurofibromas.
- Distinctive osseous lesion (Cortical thinning of long bones).
- Optic glioma.
- First degree relative with NF.
Neurofibromatosis Type 1

- Plexiform neuroma of the eyelid is strongly associated with ipsilateral optic glioma. There may be minimal visual disturbance
- Scoliosis, precocious puberty and malignant neoplasms
- Hypertension due to renal artery stenosis or pheochromocytoma

13. A 2-month-old infant is seen for a routine visit. The examination reveals a normal-looking infant except for microphallus. During the examination, he develops a generalized seizure. His blood glucose is 15 mg/dL. An intravenous bolus of D10W was given and an infusion of dextrose in water was started. The next most appropriate step is to:
   - a. Analyze the urine for 17-keto steroids
   - b. Send urine for organic and amino acid screen
   - c. Measure serum ketones
   - d. Arrange for a cranial MRI scan
   - e. Measure serum insulin and c-peptide levels

Septo-optic dysplasia

- Bilateral or unilateral optic nerve hypoplasia with absent septum pellucidum
- Variable endocrine deficiencies
- Hypoglycemia and microphallus points the diagnosis towards pan-hypopituitarism
14. A 4 year-old child presents to the emergency department with tetany. The serum calcium is 6.0mg/dL, phosphate 9.6mg/dL, and alkaline phosphatase is 586 U/dL. All of the following statements are true regarding pseudohypoparathyroidism EXCEPT:

- a. His cranial CT scan may show calcification of basal ganglia
- b. He is at risk of developing gonadal dysfunction during adult life
- c. He may have moderate learning disability
- d. The parathormone levels will be low
- e. He needs a slit lamp examination to look for cataracts

Pseudohypoparathyroidism

- Genetic defect in the hormone receptor adenylate cyclase system- unresponsiveness to the hormone
- Short stature, bradydactyly with short metacarpals
- Tetanus is the common presenting sign
- Decreased excretion of urinary phosphate and cyclic AMP after IV PTH

15. 1-month-old infant presents with constipation and poor weight gain. On examination, he has facial wasting with an inverted “V-shaped” upper lip and generalized hypotonia. The following is true regarding this infant’s condition EXCEPT:

- a. He needs an ECG to look for cardiac conduction abnormalities
- b. Serum creatine kinase levels will be markedly elevated
- c. He is at risk to develop diabetes mellitus later in life
- d. Slit lamp examination of his eyes may show cataracts
- e. He has inherited this condition from his mother
Myotonic dystrophy

- Autosomal dominant
- Multiple organ defects, progressive muscle weakness in the distal muscles
- Myotonia develops after 6 years of age
- Cataracts, cardiac conduction defects, mild to moderate mental retardation and hypogonadism
- Diabetes and adrenocortical insufficiency develops later
- CK is normal or mildly elevated