1. 5 months old infant is seen in the clinic for routine visit for immunization. Mom has concerns regarding his poor eye contact. Also he seems to startle easily. The following is true regarding his condition EXCEPT:

- a. Physical examination may show hypotonia
- b. He is at risk to develop seizure disorder
- c. He is at risk to develop severe mental retardation
- d. Specific treatment is available to halt the progression of the disease
- e. Parents need genetic counseling

Tay-Sachs disease

- No organomegaly.
- Cherry red spot: sandhoff, Nieman pick, GM1 gangliosidosis–hepatospleenomegaly
- Autosomal recessive
- Caused by excess storage of the cell membrane glycolipid, Gm2 ganglioside, within cell lysosomes
- Loss of motor skills after 5 months of age, hyperreflexia, and macrocephaly
- Decreased eye contact and increased startle response
- Relentless neurodegeneration towards death
- Diagnosis by measuring beta-hexaminidase in white blood cells and DNA testing
2. 12-year-old young lady presents with headache, nausea, vomiting, and double vision for the past 2 weeks. The following are true about her condition EXCEPT:

• a. She may have an abnormal gait
• b. Ophthalmoscopy may reveal papilledema
• c. Urgent imaging of the brain is required
• d. She has oculomotor nerve paralysis on the right side
• e. Administration of acetazolamide will reverse the condition

Oculomotor nerve palsy

• Supplies the levator muscle of the eyelid and four extraocular muscles: the medial rectus, superior rectus, inferior rectus, and inferior oblique. In addition constricts the pupil through its parasympathetic fibers that supply the smooth muscle of the ciliary body and the sphincter of the iris.
• Complete paralysis of Oculomotor nerve causes ptosis, dilation of the pupil, displacement of the eye outward and downward.
• Causes include congenital, trauma and malignancy
• Trochlear nerve (superior oblique) paralysis causes the eye to deviate upward and outward often with an associated head tilt
• Abducens nerve (lateral rectus) paralysis causes medial deviation of the eye and inability to abduct beyond midline

3. 3-years-old boy presents with recent weight gain, poor appetite, breathlessness, and abdominal discomfort. Previous week he was seen in the clinic for an URI. The following is true regarding Idiopathic nephrotic syndrome:

• a. Hypertension is common at presentation
• b. Long-term diuretic therapy is beneficial
• c. Renal biopsy is indicated to make a diagnosis in all cases
• d. Urine analysis may show microscopic hematuria in some cases
• e. Relapse is extremely rare
Idiopathic nephrotic syndrome

- Focal segmental sclerosis 10%, Mesangial proliferative 5%
- 85% of idiopathic NS are due to minimal change
- More common in boys 2-6 years
- Proteinuria, Hypoalbuminemia, edema and hyperlipidemia
- Hypertension is uncommon (10%)
- 10-20% may have microscopic hematuria
- Acute abdomen in NS: peritonitis, hypovolemia, and renal vein thrombosis
- Relapse and steroid sparing drugs: 90% responds to steroids. Cyclophosphamide, cyclosporine and mycophenolate are the second line drugs

4. 10-years-old young man with sickle cell disease presents with acute abdominal pain, fever, lethargy and cooler extremities. His diagnosis/management includes all of the following EXCEPT:

- a. He may have cholecystitis
- b. Administer a isotonic fluid bolus and continue with IV hydration
- c. His symptoms are due to acute splenic sequestration
- d. Administer broad spectrum intravenous antibiotics after obtaining appropriate cultures
- e. Administer intravenous narcotic analgesic

Sickle cell disease

- Splenic dysfunction is followed eventually by splenic infarction, usually by two to four years of age
- Altered splenic function- increased susceptibility to infections: Peritonitis, meningitis, and sepsis mainly by Pneumococci and H.influenza
- Dysfunctional IgG and IgM antibody responses, defects in alternative pathway fixation of complement, and epinephagocytic dysfunction may also play a role in the predisposition to invasive infection
- Cardiomegaly-cardiomyopathy- iron overload- heart, liver, and pancreas
- Progressive impairment in renal function due to diffuse glomerular and tubular fibrosis: Hypoalbuminuria >5years
- Zinc deficiency is prevalent and contributes to poor growth and maturation
5. 2-years-old toddler presents with failure to thrive, diarrhea, irritability and abdominal distension. The following is true regarding her condition:

- a. Sweat chloride test is diagnostic
- b. Prolonged course of metronidazole is necessary
- c. Requires gluten-free diet until adolescent growth spurt
- d. There is increased risk to develop bowel lymphoma
- e. Majority is children with this condition will develop precocious puberty

Celiac disease

- Permanent gluten sensitive enteropathy with a need for lifelong gluten free diet
- Presents between 6 months to 2 years
- Wheat, rye and barley - not oats
- Serology: Serum IgA endomyseal and transglutaminase antibodies
- Patients with a positive serology should undergo a small bowel biopsy
- Villous atrophy (short flat villi) and crypt hyperplasia
- Growth retardation, FTT, diarrhea, vomiting, muscle wasting and miserable kids
- Celiac disease is associated with a number of autoimmune disorders including type 1 diabetes mellitus and autoimmune thyroid disease.

6. This young lad with a history of asthma and eczema presents to the clinic with worsening eczema. On examination, he looks toxic, febrile (40 C), and has poor peripheral perfusion. His management includes:

- a. Referral to a dermatologist as he has intractable eczema
- b. Intravenous methylprednisolone
- c. Topical steroid and antibiotic cream and a follow-up visit in one week
- d. Hospitalization for intravenous fluids, acyclovir and broad spectrum antibiotics
- e. None of the above
7. Parents of a 10-years-old lad noticed that he has been limping for a few days. On further questioning he had no pain while walking. The following is true regarding his condition:

- a. It is almost always a unilateral condition
- b. More common in girls
- c. Affected children may have advanced bone age
- d. Surgical treatment is always necessary
- e. He is at risk to develop hip osteoarthritis without appropriate therapy

Avascular necrosis

- Legg-Calvé-Perthes disease (LCP) is a syndrome of idiopathic osteonecrosis (avascular necrosis) of the hip.
- It typically presents as hip pain and/or painless limp of acute or insidious onset in children between the ages of 3 and 12 years. It is bilateral in 10 to 20 percent of patients.
- The male-to-female ratio is 4:1 or greater, and African-Americans are rarely affected. Affected children have delayed bone age
- Diagnosis of LCP demands a high index of suspicion, because initial radiographs often are normal. Bone scan shows decreased perfusion to the femoral head, and MRI reveals marrow changes highly suggestive of the diagnosis
- Self healing, combination of surgical and non surgical management
- Prevention femoral head deformity and secondary osteoarthritis are indications for treatment. Containment of the femoral head within the acetabulum is the goal of the therapy

8. A 5 year-old has a history of malaise, fever, headache, nausea, and vomiting for the past 24 hours. He developed a rash an hour ago. Now he is lethargic, confused and combative. His pulse is 160 beats per min with a blood pressure of 70/40 mm Hg. Blood work was obtained and an intravenous line has been placed. The next most appropriate plan of action is to:

- a. Perform a lumbar puncture
- b. Obtain an urgent CT scan of the brain
- c. Intravenous mannitol to reduce ICP
- d. Administer a normal saline fluid bolus
- e. Order blood levels of ammonia
9. A 3 year-old toddler presents to you for recurrent ear infections and sinuses. He is recovering from a recent bout of bacterial pneumonia. On examination he has prominent blood vessels in the conjunctiva. The mother informs you that he has been unsteady on his feet for the past week. Appropriate management includes all of the following, EXCEPT:

- a. Measure serum immunoglobulin levels
- b. Reassure the parents that “It is all a part of growing up”
- c. Order a sweat chloride test
- d. Imaging of the brain
- e. Counsel the parents that the child may have increased risk to develop malignant tumors

Ataxia-telangiectasia

- Ataxia-telangiectasia is autosomal recessive and is associated with defective DNA repair mechanisms. Patients develop progressive cerebellar ataxia, abnormal eye movements, oculocutaneous telangiectasias, and immune deficiency.
- Telangiectasias seen primarily on the bulbar conjunctivae and on exposed areas of the skin, typically the pinnae, nose, face, and neck. In most cases, they first appear when the child reaches three to five years of age
- Immune deficiency, affecting both cellular and humoral immunity, occurs in approximately 70 percent of patients. The defect is quite variable but often manifests as recurrent sinopulmonary infections
- Absence of IgA, increased alpha feto protein, and Increased radiation-induced chromosomal breakage in cultured cells
- Also patients have other neurologic, endocrinologic, hepatic and cutaneous manifestations
- High incidence of malignancies

10. A 2 year-old presents with an abdominal mass, which was noted by the mother while bathing him. Which of the following is true regarding the abdominal mass in this child?

- a. A laxative or an enema might be needed
- b. Associated hypertension is very rare
- c. Siblings have an increased risk for similar condition
- d. A surgical option is not warranted
- e. All of the above
Wilm’s tumor

- One of the common malignancies in childhood
- Most children with Wilm’s tumor present with an abdominal mass or swelling, without other signs or symptoms. Other symptoms can include abdominal pain, hematuria, fever, and hypertension
- Once a Wilm’s tumor is suspected, subsequent abdominal examinations should be performed carefully
- Associated with genito-urinary abnormalities, hemihypertrophy, aniridia, deletions of chromosome 11 and some specific syndromes (WGAR, Drash, and BW)
- Surgery followed by chemo
- Siblings are at higher risk

11. An 8 month-old infant is seen with a 3-week history of diarrhea, abdominal distension, rectal prolapse, and poor weight gain. Past medical history is significant for hospitalization for staphylococcal pneumonia at the age of 3 months. The next most appropriate plan of action is to:

- a. Obtain an urgent surgical evaluation
- b. Obtain serum immunoglobulin levels
- c. Obtain serum anti-gliadin antibodies
- d. Change her milk formula to a soy-based formula
- e. Perform a sweat chloride test

Cystic fibrosis

- 1 in 3500 Caucasian live births and 1 in 17000 African American infants in the US
- AR, gene codes for the CFTR expressed in epithelial cells of airways, GI tract, sweat glands, and GU system.
- Common presentation: Respiratory infections, loose stools, failure to thrive, malnutrition, meconium ileus and intestinal obstruction
- Uncommon presentation: Electrolyte abnormality, nasal polyps, rectal prolapse, hepatobiliary disease, hyproproteinemia, and azoospermia
Cystic fibrosis

- Diagnostic criteria: Presence of typical clinical features or history of CF in a sibling or positive newborn screening PLUS
- Lab evidence of CFTR dysfunction: two elevated sweat chloride on separate days or identification of two CF mutations or abnormal nasal potential difference measurement
- Abnormal sweat test: adrenal insufficiency, ectodermal dysplasia, hereditary nephrogenic DI, G6PD, hypothyroidism, hypoparathyroidism etc

12. The parents of this 6-month-old infant are concerned about sudden jerking movements of the whole body. She seems slow to catch up on her milestones. She is not smiling and not able to roll over yet. All of the following is true about her EXCEPT:

- a. She needs evaluation for seizure disorder
- b. She needs evaluation of her heart with ECHO
- c. She may develop renal cysts
- d. She has inherited this condition from her mother
- e. Her future siblings have higher risk for the same condition

Tuberous sclerosis

- Autosomal dominant: 1 in 6000, 50% of cases are new mutations
- Heterogeneous disease with wide clinical spectrum
- Many organ systems are affected.
- Tubers in the brain, hypsarrhythmia, rhabdomyomas of the heart (50%), hamartomas or polycystic kidneys, and angiomyolipomas in the lungs lead to spontaneous pneumothorax