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

• Complete paralysis of Oculomotor nerve causes ptosis, dilation of the pupil, displacement of the eye outward and downward.
• 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 and cyclophosphamide is second line drug

Sickle cell disease

- Altered spleenic function: increased susceptibility to infections: Peritonitis, meningitis, and sepsis mainly by Pneumococci and H.influenza
- Also have deficient levels of opsonins: H.influenza, Salmonella and Pneumococcus
- Cardiomegaly-cardiomyopathy: iron over load- heart, liver, and pancreas
- Progressive impairment in renal function due to diffuse glomerular and tubular fibrosis: Hypostenuria >5years
- Zinc deficiency is prevalent and contributes to poor growth and maturation
5. A 2-year-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

- Gluten sensitive enteropathy—permanent
- Presents between 6 months to 2 years
- Wheat, rye and barley— not oats
- Villous atrophy (short flat villi) and crypt hyperplasia
- Serum IgA endomyseal antibody test (almost 100% sensitive and specific)
- Antibody to transglutaminase antigen
- Initial small bowel biopsy probably still needed
- Growth retardation, FTT, diarrhea, vomiting, muscle wasting and miserable kids
- Lifelong gluten free diet

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 should include:

- a. Referral to a dermatologist as he has intractable eczema
- b. Intravenous methylprednisolone
- c. Topical steroid and antibiotic cream and arrange for a follow-up visit the next week
- d. Hospitalizing him for intravenous fluids, acyclovir and 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

- Primary disorder of males, classic presentation of painless limp
- Interruption of blood supply to capital femoral epiphysis
- Associated with deficiency of protein C, S
- Bilateral in 20%
- Affected children have delayed bone age
- Self-healing and 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 sinusitis. 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. Inform the parents that the child has increased risk to develop malignant tumors

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Ataxia-telangiectasia

• Neurologic, immunologic, endocrinologic, hepatic and cutaneous manifestations
• Progressive cerebellar ataxia/chronic sino pulmonary disease
• High incidence of malignancies and defective DNA repair
• Absence of IgA and decreased IgE

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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

- Common malignancy in childhood
- Associated with genito-urinary abnormalities/hemihypertrophy/aniridia/deletions of chromosome 11
- Hypertension in 60%
- 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 white live births and 1 in 17000 black 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, hydropneumonia, and azoosperma
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

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:

- She needs evaluation for seizure disorder
- She is at risk to develop heart failure or arrhythmias
- She may develop renal cysts
- She has inherited this condition from her mother
- 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