Case 1

- Patient AB is a 4 year old previously healthy male who presents to your clinic with intermittent episodes of coughing, fast breathing, and chest tightness. These episodes seem to be worse when he gets colds and are worse around his uncle who smokes. He once tried a friend’s “inhaler” which seemed to help his symptoms.

- What is the most likely diagnosis?
Asthma
Definition and Epidemiology

- Asthma is a chronic, inflammatory lung disease characterized by the following:
  - Symptoms of cough, wheezing, dyspnea, and chest tightness that occur in paroxysms and are usually related to specific triggering events
  - Airway narrowing that is partially or completely reversible
  - Increased airways responsiveness to a variety of stimuli
- Prevalence = 9.6% in children ≤ 18

Asthma
Risk Factors and Triggers

- Risk Factors
  - Boys, non-white ethnicity, lower socioeconomic status, and urban inhabitants are more likely to have asthma
- Triggers
  - Inhaled allergens, respiratory infections, inhaled respiratory irritants, hormonal fluctuations, medications, exercise, weather, emotional disturbance


Asthma
Pathophysiology

- Symptoms are due to airflow obstruction caused by the following:
  - Smooth muscle constriction around airways
  - Airway wall edema
  - Intraluminal mucus accumulation
  - Inflammatory cell infiltration of the submucosa
  - Basement membrane thickening


Asthma
Clinical Presentation

- Signs of respiratory distress
  - Examples: tachypnea, belly breathing, retractions, nasal flaring
  - Hypoxia
  - Wheezing, decreased breath sounds
  - Prolonged inspiratory to expiratory ratio

Case 1 Continued

- With further history, you learn that AB experiences his symptoms 3-4x per week and even wakes up from sleep 1x per week with coughing.

- What is his classification of asthma severity?
  - A) Intermittent
  - B) Mild persistent
  - C) Moderate persistent
  - D) Severe persistent

- Based on asthma severity classifications, what is his FEV1 likely to be?
  - A) >80% predicted
  - B) 60-80% predicted
  - C) <60% predicted

- What type of controller medication would you prescribe?
  - A) None
  - B) Inhaled steroid
  - C) Long-acting bronchodilator
  - D) Leukotriene inhibitor

---

### Asthma Classification and Treatment

<table>
<thead>
<tr>
<th>Severity</th>
<th>Symptoms</th>
<th>Nighttime Symptoms</th>
<th>Exacerbations</th>
<th>Lung Function (FEV1 or PEF)</th>
<th>Maintenance Therapy</th>
</tr>
</thead>
<tbody>
<tr>
<td>Intermittent</td>
<td>&lt; 2x/week</td>
<td>&lt; 2 / month</td>
<td>Occasional</td>
<td>&gt;80% predicted</td>
<td>None</td>
</tr>
<tr>
<td>Mild Persistent</td>
<td>&gt; 2x/week</td>
<td>&gt; 2 / month</td>
<td>&gt; 2x / week</td>
<td>&gt;80% predicted</td>
<td>Inhaled steroid</td>
</tr>
<tr>
<td></td>
<td>&lt; Daily</td>
<td>&gt; 2 / month</td>
<td></td>
<td></td>
<td>Leukotriene inhibitor</td>
</tr>
<tr>
<td>Moderate</td>
<td>Daily</td>
<td>&gt;1 / week</td>
<td>&gt; 2x / week</td>
<td>60%-80% predicted</td>
<td>Inhaled steroid</td>
</tr>
<tr>
<td>Persistent</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Long-acting bronchodilator</td>
</tr>
<tr>
<td>Severe</td>
<td>Continual</td>
<td>Frequent</td>
<td>Frequent</td>
<td>&lt;60% predicted</td>
<td>Inhaled steroid</td>
</tr>
<tr>
<td>Persistent</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Leukotriene inhibitor</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Oral steroid</td>
</tr>
</tbody>
</table>

### Asthma Acute Exacerbation Treatment

- **Albuterol (if mild) or albuterol + ipratropium bromide**
  - Systemic steroid therapy if not improved after the first inhaled treatment
- **Supplemental oxygen for saturations <92%**
  - **Terbutaline**
  - **Magnesium Sulfate**
  - **Epinephrine**
Other Atopic Conditions

- Include asthma, atopic dermatitis, and allergic rhinitis

Exposure to allergen (ex dust, pollens, animal dander, medications, foods, insect bites)

Production of IgE and eosinophils

Release of allergic mediators from mast cells and basophils

Case 1 Continued

- Once AB has one atopic condition, how much more likely is he to develop a second atopic condition as compared to an unaffected child?

  - A) Equally as likely
  - B) 2x as likely
  - C) 3x as likely
  - D) 5x as likely

Eczema (Atopic Dermatitis)

Definition and Epidemiology

- Definition
  - Chronic, relapsing dermatosis with features of dry skin and pruritus
  - Associated with a personal or family history of atopic conditions

- Epidemiology
  - In the United States, 15%-20% of children are diagnosed with atopic dermatitis
  - Atopic Triad = Atopic dermatitis, asthma, allergic rhinitis
  - Children who have one component are 3x as likely to develop a second component
Eczema (Atopic Dermatitis)
Pathophysiology and Clinical Presentation

- Pathophysiology
  - Impaired epidermal barrier structure and function
  - Inflammatory response triggered by environmental factors

- Clinical Presentation
  - Primary Changes:
    - Erythematous, scaly, poorly defined papules / plaques
  - Secondary Changes:
    - Pigmentation changes, lichenification
    - Infection may be present
  - Distribution:
    - Infants: cheeks, dorsa of wrist, ankles, lateral extremities
    - Children: flexural involvement, neck, antecubital and popliteal fossae, gluteal folds
    - Teens: Eyelids, hands, and feet

Allergic Rhinitis
Definition and Epidemiology

- Definition:
  - Inflammation of the membrane lining of the nose due to an IgE mediated hypersensitivity reaction to specific allergens

- Epidemiology:
  - 42% of children are diagnosed with allergic rhinitis by the age of 6yo

Allergic Rhinitis
Clinical Presentation

- Symptoms:
  - Nose: congestion, rhinorrhea, itching, sniffing, snorting
  - Other: Mouth breathing, snoring, nasal voice, cough (usually secondary to postnasal drainage), itchiness of eyes and throat

- Physical Exam:
  - Nose: Obstruction, pale mucosa, enlarged/boggy turbinates, clear secretions
  - Other: Allergic shiners, allergic crease, pharyngeal cobblestoning
Allergic Rhinitis
Management
- Allergen Avoidance
  - Includes decreasing exposure to pets, pollen, mold, dust mites, smoke
- Pharmacotherapy
  - Corticosteroids (intranasal)
  - Antihistamines (oral, intranasal)
  - Leukotriene inhibitors
  - Decongestants (only intermittently)

Case 2
- CD is a newborn who had a healthy delivery. Her post-delivery hospitalization was only notable for first passage of meconium at 48 hours. One week after delivery, you are called with abnormal newborn screen results.

- What diagnosis are you worried about?

Case 2 Continued
- If CD does have this diagnosis which of the following is not an associated complication?

  - A) Failure to thrive
  - B) Decreased fertility
  - C) Focal biliary cirrhosis
  - D) Neutropenia

Cystic Fibrosis
Cystic Fibrosis
Definition, Epidemiology, and Genetics

- **Definition**
  - Disorder of an exocrine membrane chloride channel (cystic fibrosis transmembrane conductance regulator or CFTR protein) resulting in pulmonary, gastrointestinal, and reproductive dysfunction

- **Epidemiology**
  - In Caucasians, affects 1:3000 births, 1:25 is a carrier
  - Prevalence in non-Caucasians is rising

- **Genetics**
  - Autosomal recessive mutations, chromosome 7

Case 2 Continued

- CD’s (much) older brother is planning to marry a Caucasian person from the general population. What are the odds of the two of them having a child with cystic fibrosis?
  - A) 1/150
  - B) 1/200
  - C) 1/25
  - D) 1/100

  \[
  (2/3 \text{ chance of sibling carrier}) \times (1/25 \text{ chance in Caucasian population}) \times (1/4 \text{ chance of child having two recessive genes}) = 1/150
  \]

Cystic Fibrosis
Clinical Manifestations - Pulmonary

- **Symptoms:**
  - Persistent, productive cough
  - Increase thick sputum production, tachypnea, dyspnea

- **Physical Exam:**
  - Increased anterior to posterior chest diameter, digital clubbing, crackles, increased inspiratory to expiratory ratio

- **Diagnostics:**
  - Hyperinflation on chest radiography
  - Obstructive pattern on pulmonary testing

Cystic Fibrosis
Clinical Manifestations - Gastrointestinal

- **Pancreatic Disease**
  - Exocrine dysfunction
    - Insufficient secretion of digestive enzymes -> malabsorption of fat, protein -> steatorrhea, failure to thrive, vitamin deficiency
  - Endocrine dysfunction
    - CF-associated diabetes
    - Pancreatitis

- **Intestinal obstruction**
  - Delayed passage of meconium / meconium ileus
  - Distal intestinal obstructive syndrome

- **Biliary Disease**
  - Focal biliary cirrhosis due to inspissated bile
  - Cholelithiasis due to loss of bile acids and consequent production of lithogenic bile
### Cystic Fibrosis

#### Clinical Manifestations – Reproductive, Musculoskeletal, and Other

- **Reproductive dysfunction**
  - Men: 95% are infertile due to defects in sperm transport
  - Women: 20% are infertile due to secondary amenorrhea from malnutrition and from abnormal cervical mucus

- **Musculoskeletal**
  - Reduced bone mineral content secondary to malnutrition
  - Hypertrophic osteoarthropathy
  - Arthropathy

- **Other**
  - Recurrent venous thrombosis
  - Nephrolithiasis, nephrocalcinosis

*Hypertrophic osteoarthropathy.* 

### Cystic Fibrosis

#### Diagnosis

- **Indications for testing:**
  - Clinical symptoms suggestive of cystic fibrosis, positive newborn screen, or a sibling with cystic fibrosis

- **Diagnosis:**
  - Clinical symptoms consistent with CF in at least one organ system
  - Elevated sweat chloride >60 mmol/L

- **Other testing**
  - Presence of two disease-causing mutations in CFTR can aid in or confirm a diagnosis
  - Abnormal nasal potential difference can be used if other testing is inconclusive

### Cystic Fibrosis

#### Treatment

- **General**
  - Close following of growth, nutritional status, psychosocial situation

- **Pulmonary**
  - Obstruction: Bronchodilators
  - Colonization: Antibiotics including tobramycin for pseudomonas aeruginosa colonization and azithromycin for both its antibiotic and anti-inflammatory effects
  - Inflammation: Steroids, azithromycin
  - Mucus clearance: Inhaled DNAase and hypertonic saline

- **Gastrointestinal**
  - Pancreatic insufficiency: Pancreatic enzyme replacement therapy
  - Obstruction: Cathartics, enemas

### Case 2 Continued

- You follow CD throughout her childhood into her teenage years. At age 17, she presents with poor weight gain and you decide you want to evaluate her for diabetes. All of the following lab values give you a diagnosis of diabetes except:

A) Fasting plasma glucose ≥126 mg/dL
B) Symptoms of hyperglycemia + random venous glucose ≥300 mg/dL
C) Abnormal glucose tolerance test (A plasma glucose ≥200 mg/dL measured two hours after a glucose load of 1.75 g/kg)
D) Glycated hemoglobin (A1C) ≥ 6.5 percent
Diabetes Mellitus

Definitions

• Definition: Disorder of metabolic homeostasis controlled by insulin resulting in abnormal carbohydrate and lipid metabolism

• Type 1: Destruction of islet cells -> insulin deficiency

• Type 2: Insulin resistance -> relative insulin deficiency

Clinical Presentations

• Classic new onset
  • Polyuria, polydipsia, weight loss

• Diabetic Ketoacidosis
  • Classic symptoms with signs of acidosis including abdominal pain, nausea/vomiting, hyperventilation with Kussmaul breathing, drowsiness / lethargy / coma

• Asymptomatic

Diabetes Mellitus

Diagnosis

• Diabetes Mellitus:
  • Fasting plasma glucose ≥126 mg/dL
  • Symptoms of hyperglycemia + random venous plasma glucose ≥200 mg/dL
  • Abnormal glucose tolerance test
    • A plasma glucose ≥200 mg/dL measured two hours after a glucose load of 1.75 g/kg
  • Glycated hemoglobin (A1C) ≥ 6.5 percent

• Diabetic Ketoacidosis
  • Blood glucose of > 200mg/dL
  • Metabolic acidosis (venous pH <7.3 and/or plasma bicarbonate <15 mEq/L)
  • Hyperketosis and hyperosmolality

Diabetes Mellitus
Diagnosis – When to screen for Type 2

- **Overweight**
  - Body mass index >85th percentile for age and sex
  - Weight-for-height >85th percentile
  - Weight >120% of ideal for height

- **Two other risk factors**
  - Family history of type 2 diabetes
  - High-risk racial/ethnic background
  - Presence of a condition associated with insulin resistance
    - Acanthosis nigricans, polycystic ovarian syndrome, hypertension, or dyslipidemia
  - Maternal history of gestational diabetes or diabetes

Diabetes Mellitus
Diagnosis – Distinguishing type 1 and type 2

<table>
<thead>
<tr>
<th></th>
<th>Type 1</th>
<th>Type 2</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Body habitus</strong></td>
<td>Normal weight, recent history of weight loss</td>
<td>Overweight, obese</td>
</tr>
<tr>
<td><strong>Age</strong></td>
<td>Peaks in incidence &lt;14yo</td>
<td>Onset after puberty</td>
</tr>
<tr>
<td><strong>Signs of insulin resistance</strong></td>
<td>None</td>
<td>Acanthosis nigricans, hypertension, dyslipidemia, PCOS</td>
</tr>
<tr>
<td><strong>Family history</strong></td>
<td>Close relative with type 1 DM</td>
<td>Close relative with type 2 DM</td>
</tr>
<tr>
<td><strong>Labs consistent with autoimmune disease</strong></td>
<td>Presence of islet-specific pancreatic autoantibodies</td>
<td>None</td>
</tr>
</tbody>
</table>

Diabetes Mellitus
Treatment

- **Type 1**
  - Insulin
    - Conventional Regimen:
      - Intermediate-acting BID (2/3 at breakfast, 1/3 at dinner)
      - Short-acting 2-3x per day
    - Intensive Regimen:
      - Long-acting insulin (basal)
      - Short-acting 3x per day with meals
      - Insulin Pump

- **Type 2**
  - Lifestyle modifications including exercise, maintaining a healthy diet, and weight loss
  - Metformin

Diabetes Mellitus
Complications and Screening

- Associated autoimmune disease
  - Thyroid dysfunction – TSH at diagnosis, every 1-2 years, and if symptoms
  - Celiac disease – IgA / TTG at diagnosis and if symptoms

- Growth disturbance
  - Physical exam during each visit

- Retinopathy
  - Ophthalmology screening starting at 10 years old and after patient has had DM for 3-5 years, with follow-up every year

- Nephropathy
  - Urine microalbumin screen every year, blood pressure checks at each visit

- Neuropathy
  - Physical exam during each visit

- Microvascular / Lipids
  - Fasting lipid panel at diagnosis if there is a concerning family history or starting at age 12 if no concerning family history
Case 3

EF is a 6 year old male who has recently immigrated from Africa. He presents with left-sided weakness and slurred speech. On history, you learn that he has had multiple episodes of acute pain that took days to resolve and he has received two transfusions. How would you diagnosis his underlying disorder?

- A) Head CT with IV contrast
- B) Head MRI
- C) Hemoglobin electrophoresis
- D) Basic labs including a CBC, BMP, and inflammatory markers

Sickle Cell Disease

Definition and Epidemiology

- Definition
  - A group of heterogeneous disorders affecting hemoglobin
  - Specifically, a mutation causes hydrophilic glutamic acid to be substituted for a hydrophilic valine on the β globin gene
  - This result is an unstable form of hemoglobin that easily adapts a sickled shape

- Epidemiology (in the United States):
  - Sickle hemoglobinopathies: 90,000-100,000 people
  - Sickle cell trait is carried by 7%-8% of people with African ancestry

Sickle Cell Disease

Clinical Manifestations / Treatment

- Effects on the blood
  - Anemia
  - Mild thrombocytosis and/or leukocytosis

- Infection
  - Streptococcus pneumonia infection:
    - Due to functional asplenia
  - Prophylaxis with PCV 13 vaccination and daily penicillin
  - Fevers need urgent medical evaluation, empiric antipneumococcal antibiotics, possible hospitalization
  - Parvovirus B19 infection
    - Can lead to an aplastic crisis and resultant severe anemia
    - Treat with transfusions until bone marrow function resumes

Sickle Cell Disease
Clinical Manifestations / Treatment - Continued

- **Vaso-Occlusive Crisis**
  - Acute bone pain, lasts minutes to days
  - Pathophysiology: Bone marrow ischemia with resultant infarction
  - Triggers: Infection, dehydration, emotional stress, high altitude
  - Treatment: Opioid analgesics, anti-inflammatory medications, intravenous fluids

- **Dactylitis**
  - Specific type of vaso-occlusive crisis that occurs in infants and young children
  - Defined by tender, erythematous, and edematous hands or feet
  - Treatment: analgesics, Intravenous fluids

- **Pulmonary Complications**
  - **Acute Chest Syndrome**
    - Defined as a new pulmonary infiltrate on chest radiograph with fever, tachypnea, dyspnea, hypoxia, and/or chest pain
    - Causes include unknown etiology, infection, pulmonary infarction, fat embolism
    - Treatment includes supplemental oxygen, empiric antibiotics, bronchodilators, analgesics, intravenous fluids, transfusion, corticosteroids
  - **Asthma**
    - Associated with higher rates of acute chest syndrome, vaso-occlusive crisis, and early death
  - **Pulmonary Artery Hypertension**

- **Neurologic Manifestations**
  - **Stroke**
    - May have symptoms or be silent in nature
    - Treat with pRBC transfusion or partial exchange transfusion to reduce sickled hemoglobin
    - Screening annually with transcranial doppler ultrasonography

- **Other clinical manifestation**
  - Splenic sequestration
  - Priapism
  - Cholelithiasis

- **Treatments**
  - Hydroxyurea
    - Increases expression of fetal Hg
  - Chronic Transfusion Therapy
    - Used in stroke prevention but may also help with pain and growth
    - Complicated by iron overload, treated with chelation therapy
  - Hematopoietic Bone Marrow Transplantation

---

Sickle Cell Disease
Clinical Manifestations / Treatment - Continued

Case 4

- GH is a newborn with clinical features consistent with Down Syndrome. What is the most common abnormality seen in a Down Syndrome patient on physical exam at birth?
  - A) Murmur
  - B) Facial dysmorphism
  - C) Hypotonia
  - D) Brushfield spots
Down Syndrome

Definition
• The most common chromosome abnormality among liveborn infants
• Underlying genetic defect is trisomy 21

Dysmorphic Features
• Head and Neck:
  - UpSlanting palpebral fissures, epicanthic folds, flat facial profile/flat nasal bridge, folded or dysplastic ears, low-set small ears, brachycephaly, brushfield spots, protruding tongue, short neck, excessive skin at nape of the neck
• Extremities:
  - Short broad hands, incurved fifth finger with hypoplastic mid phalanx, transverse palmar crease, space between the first and second toes, hyperflexibility of joints

Clinical Manifestations and Screening

<table>
<thead>
<tr>
<th></th>
<th>Clinical Manifestations</th>
<th>Screening Recommendations</th>
</tr>
</thead>
<tbody>
<tr>
<td>Growth</td>
<td>Obesity, growth disturbances secondary to other disorders</td>
<td>Evaluation at health maintenance visits</td>
</tr>
<tr>
<td>Cardiac</td>
<td>Congenital heart disease (ASD, VSD, AV Canal), mitral valve prolapse, aortic regurgitation</td>
<td>ECHO at birth, cardiology follow-up as teenagers</td>
</tr>
<tr>
<td>Gastrointestinal</td>
<td>Duodenal atresia/stenosis, imperforate anus, esophageal atresia, tracheoesophageal fistula, hirschsprung disease, celiac disease</td>
<td>Symptom based evaluation</td>
</tr>
</tbody>
</table>

### Down Syndrome

**Clinical Manifestations and Screening - Continued**

<table>
<thead>
<tr>
<th>Clinical Manifestations</th>
<th>Screening Recommendations</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hearing</td>
<td>Senorineural hearing loss</td>
</tr>
<tr>
<td>Ophthalmologic</td>
<td>Strabismus, nystagmus, cataracts, refractive errors</td>
</tr>
<tr>
<td>Thyroid Function</td>
<td>Both hypothyroidism and hyperthyroidism are common</td>
</tr>
<tr>
<td>Hematology</td>
<td>Myeloproliferative disorder, polycythemia, anemia, leukemia</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Clinical Manifestations</th>
<th>Screening Recommendations</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pulmonary</td>
<td>Sleep Apnea</td>
</tr>
<tr>
<td>Neurologic</td>
<td>Dementia (early onset)</td>
</tr>
<tr>
<td>Musculoskeletal</td>
<td>Atlantoaxial instability</td>
</tr>
<tr>
<td>Dental / Periodontal</td>
<td>Periodontal disease, including inflammation, periods of acute infection, and pain</td>
</tr>
<tr>
<td>Reproduction</td>
<td>Decreased fertility, Risk of sexual abuse</td>
</tr>
</tbody>
</table>

---

### Cerebral Palsy

**Definition and Epidemiology**

- **Definition**
  - Heterogeneous group of static encephalopathy characterized by motor and postural dysfunction early in life

- **Epidemiology**
  - Prevalence is 3.6 out of 1000 children
Case 5

- Patient LJ presents in preterm labor and while you are counseling her, she asks you about cerebral palsy. What is the most common risk factor for cerebral palsy in premature newborn?
  - A) Perinatal asphyxia / Neonatal encephalopathy
  - B) Prematurity
  - C) Pre-Eclampsia
  - D) Perinatal infection

Cerebral Palsy

Risk Factors
- Perinatal asphyxia, Neonatal encephalopathy
- Stroke, intracranial hemorrhage
- Congenital abnormalities
- Multiple births
- Intrauterine infection
- Prematurity
- Kernicterus

Clinical Classification

<table>
<thead>
<tr>
<th>Type</th>
<th>Subtype</th>
<th>Clinical Presentation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Spastic</td>
<td>Diplegia</td>
<td>Lower limbs affected, walking is delayed, tip-toe walking, normal cognitive function</td>
</tr>
<tr>
<td></td>
<td>Hemiplegia</td>
<td>Affects one side of body, must occur before the age of two to be considered CP, 25% are cognitively impaired</td>
</tr>
<tr>
<td></td>
<td>Quadriplegia</td>
<td>Affects all four extremities, lower-upper, more likely to be associated with intellectual disability, seizures, feeding difficulties, speech difficulties, visual disturbance</td>
</tr>
<tr>
<td>Dysskinetic</td>
<td>Athetosis</td>
<td>Slow, smooth, writhing movements of distal muscles</td>
</tr>
<tr>
<td></td>
<td>Chorea</td>
<td>Rapid, irregular, unpredictable contractions of individual muscles</td>
</tr>
<tr>
<td></td>
<td>Dystonia</td>
<td>Repetitive, patterned, twisting, sustained movements, affects all four limbs and trunk</td>
</tr>
<tr>
<td>Ataxic</td>
<td>Atonic</td>
<td>Incoordination, congenital hypotonia, associated with motor/language delay</td>
</tr>
<tr>
<td></td>
<td>Atonic</td>
<td>Severe hypotonia, retention of primitive reflexes, may not walk</td>
</tr>
</tbody>
</table>

Management

- Requires a multidisciplinary team
  - Focus on medical, social, psychological, educational, and therapeutic needs
- Spasticity treatments
  - Botulinum toxin, oral antispastic drugs (dantrolene, benodiazepine, baclofen), intrathecal treatment (baclofen), physical therapy
- Contracture treatments
  - Physical therapy, muscle-tendon release surgery
Case 6

- KL is a previously healthy 3 year old who presents an episode of unconscious shaking in the context of a fever and upper respiratory infection.

- What is the most likely diagnosis?

- KL's parents want to know how likely he is to have a repeat episode. All of the following are risk factors for recurrence except:
  - A) High fever at the time of first seizure
  - B) Young age
  - C) Family history of febrile seizures
  - D) Short time between fever onset and seizure

Seizure Disorders

Generalized Seizures

- Generalized Seizures
  - Generalized tonic-clonic seizures
  - Febrile seizures
  - Absence seizures

- Partial Seizures
  - Simple partial seizures
  - Complex partial seizures
  - Status Epilepticus

Febrile Seizures

- Epidemiology:
  - Peak age = 8-20 months; Prevalence = 3% of children

- Classification:
  - Simple: < 15 minutes, has no focal component
  - Complex: > 15 minutes, has a focal component, recurs within 24 hours

- Presentation:
  - Associated with a rapid rate of rise in temperature
  - Commonly seen with upper respiratory tract infections and acute gastroenteritis
Generalized Seizures  
Febrile Seizures - Continued

- **Treatment:**
  - Reassurance
  - Tylenol or ibuprofen for fever control
  - Antiepileptic medications are rarely needed

- **Recurrence:**
  - 1/3 of children who have one febrile seizure will have a second
  - Risk factors for recurrence include 1) low fever at the time of first seizure, 2) young age, 3) family history of febrile seizures, 4) a short time between fever onset and seizure
  - Children who have a single febrile seizure have twice the rate of epilepsy as the general population (1% vs 0.5%)

---

Generalized Seizures  
Absence Seizures

- **Presentation:**
  - Brief staring spells, during which time the child is unresponsive
  - No postictal period

- **Diagnosis:**
  - 3 per second spike and wave pattern on EEG
  - Hyperventilating can induce

- **Treatment:**
  - Ethosuximide or valproic acid

---

Generalized Seizures  
Tonic-Clonic Seizures

- **Presentation:**
  - Tonic Phase:
    - Flexion of trunk, and extension of back, arms, and legs typically lasting 30 seconds
  - Clonic Phase:
    - Convulsive movements / tremors alternating with atonia during which time the child may be void, typically lasting 1-2 minutes
  - Postictal Phase:
    - Initially child is unconscious with gradual awakening and orientation

- **Treatment:**
  - Valproic acid, phenytoin, carbamazepine, lamictal

---

Partial Seizures  
Simple Partial Seizures

- **Presentation:**
  - Typically motor activity in an extremity, though may occur as sensory seizures or autonomic seizures as well
  - Consciousness is maintained
  - Often occur wen the patient falls asleep or just wakes up

- **Treatment:**
  - Carbamazepine, though treatment may not always be necessary

- **Prognosis:**
  - Some cases remit during adolescence
Partial Seizures
Complex Partial Seizures
- Presentation:
  - Commonly include facial movements, automatisms
  - Impaired consciousness
  - May be difficult to distinguish from absence seizures

- Treatment:
  - Valproic acid, phenytoin, carbamazepine, lamictal

Seizure Disorders
Status Epilepticus
- Definition:
  - Continued seizure activity for 30 minutes or recurrent seizures without return to baseline

- Treatment:
  - Benzodiazepines (typically lorazepam, repeated up to 3x every 5 minutes)
  - Load anticonvulsive agent (typically fosphenytoin)
  - Consider phenobarbital, midazolam, or propofol for refractory seizures

References
- Asthma
References

• Allergic Rhinitis

• Atopic Dermatitis

• Cystic Fibrosis

• Diabetes Mellitus

• Sickle Cell Disease

• Down Syndrome

• Seizure Disorders