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# Genetic Disorders

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Bio 156

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# Autism Spectrum Disorder

## What is it?

- It is borderline a genetic disorder
- “a developmental disability caused by differences in the brain” (CDC)
- “Problems with social communication and interaction” (CDC)
- “Restricted or repetitive behaviors or interests” (CDC)
- “ 39% of people had a change in one of the two copies of the HOXA1 gene, which is located on Chromosome 7” (Magellan Healthcare)

## Signs and symptoms:

- Delayed language skills
- Delayed movement skills
- Delayed cognitive or learning skills
- Avoids or does not keep eye contact
- Does not respond to name by 9 months of age



# Autism Spectrum Disorder

## Treatment and Interventions:

- Applied Behavior Analysis (ABA): encourages desired behaviors and discourages undesired behaviors to improve a variety of skills.
- Occupational Therapy: teaches skills that help the person live as independently as possible
- Treatment and Education of Autistic and Related Communication-Handicapped Children (TEACCH): provides teachers with ways to adjust the classroom and improve academic and other outcomes.
- Cognitive Behavioral Therapy (CBT): Focuses on learning the connections between thoughts, feelings, and behaviors.
- There is no official cure for autism but, these treatments can help

# Cystic Fibrosis (CF)

## What is it?

- "An autosomal recessive disease caused by mutations of a gene located on the long arm of chromosome 7(1)." (Stern)
- Common life-shortening disease that afflicts people of Northern European descent. (Zheng)
- Affects Respiratory tract, Sinus, digestive tract, reproductive system, and growth disorder. (Zheng)

## Signs and Symptoms

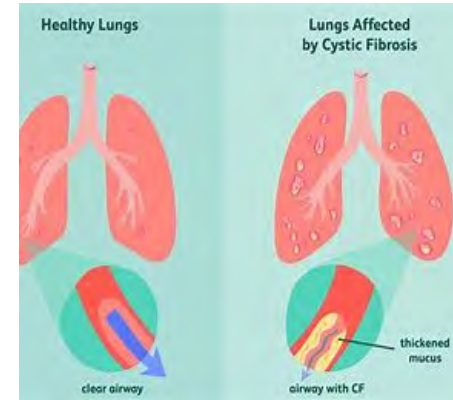
- Persistent Cough with Phlegm
- Postnasal Drip
- Thickened mucus, sweat, and digestive fluids.
- Male Infertility
- Diarrhea/Constipation
- Repeated Lung Infections and damaged airways.



# Cystic Fibrosis (CF)

## Treatments

- Diagnosis starts with sweat chloride test used for CF diagnosis (Zheng)
- Mucus thinner
- Airway clearance
- Antibiotics
- Inhalation Therapy (Hypertonic saline to hydrate thick mucus)
- Long-term azithromycin.
- Pancreatic enzymes.
- Lung transplant (Zheng)
- Staying away from other CF patients to no cross germs.



There is no cure and CF can reduce life expectancy by more than half. But increased research over the years has improved life expectancy to age 40. (Zheng)

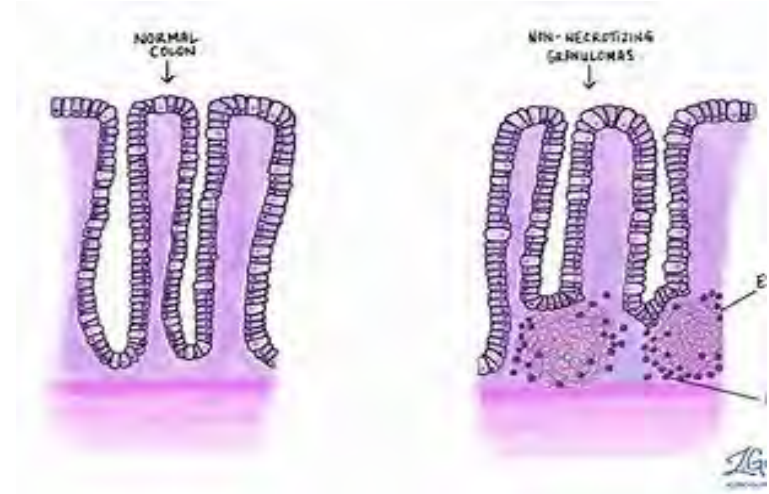
# Crohn's Disease

## What is it?

- Relapsing systemic inflammatory disease (Baumgart)
- Can affect any part of the gastrointestinal tract (mouth to anus).
- "Genome wide association studies and computerised meta-analyses have identified and confirmed 71 susceptibility loci for Crohn's disease on 17 chromosomes so far," (Baumgart)

## Signs and Symptoms (Baumgart)

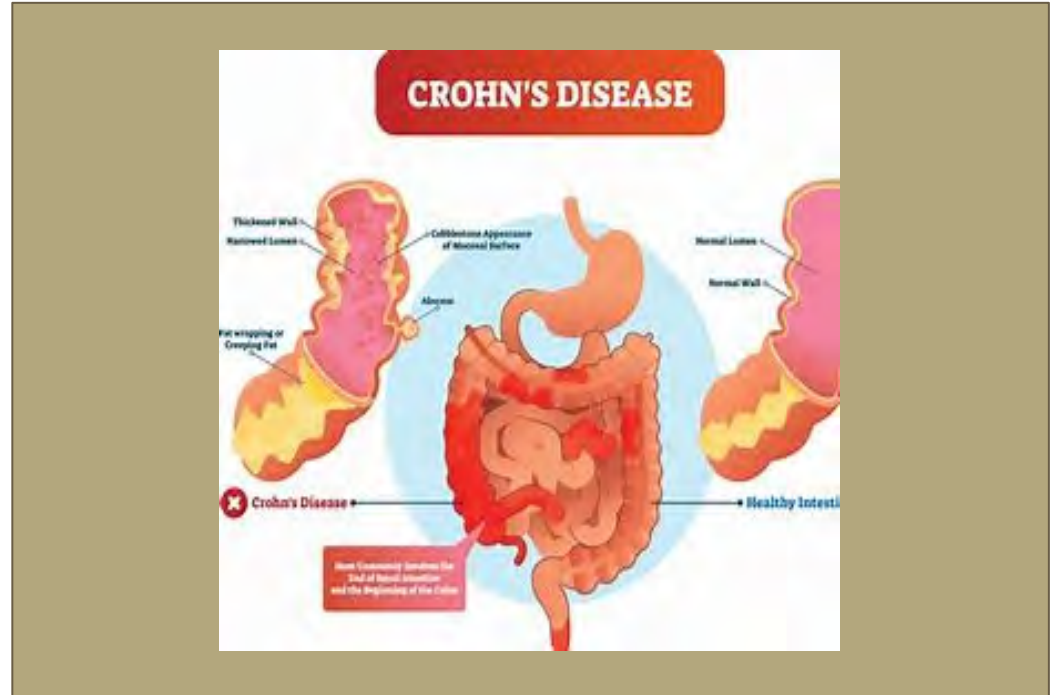
- Blood or mucus in stool
- Abdominal pain
- Diarrhea
- Fatigue
- Weight Loss
- Malnutrition



# Crohn's Disease

## Treatments (Baumgart)

- Full ileocolonoscopy
- Avoid Smoking
- Steroids
- Immunosuppressants
- Biologics
- Antibiotics
- Dietary change



# Turner Syndrome

## Genetic Abnormality:

- "Turner's syndrome may be defined as the combination of characteristic physical features and complete or part absence of one of the X chromosomes, frequently accompanied by cell-line mosaicism." (Ranke)

## Signs and Symptoms:

- Reduced height
- low hairline
- flat feet
- broad chest
- lazy eye
- webbed neck,
- receding lower jaw
- infertility.
- "Morbidity and Mortality are increased."(Gravholt)



# Turner Syndrome

## Treatments:

- Gonadal Dysgenesis allows a gain in adult height. (long-term consequences are not clear)
- Estrogen Replacement Therapy
- Growth Hormone Therapy



# Sickle Cell Disease

## What is it?

- Sickle Cell Disease is a collection of inherited red blood cell disorders that result in defective hemoglobins and make red blood cells stiff, sticky, and resembling the "Sickle" farm tool.
- The gene that is affected in a person with SCD is the HBB gene which is supposed to provide instructions to make beta-globin, a protein that is a part of the hemoglobin. The hemoglobin is a protein in red blood cells that carry oxygen throughout the body.
- "SCD is a genetic condition that is present at birth. It is inherited when a child receives two genes—one from each parent—that code for abnormal hemoglobin." (CDC)

# Sickle Cell Disease

## Signs and Symptoms:

### Babies/Toddlers

- A yellowish color of the skin (Jaundice)
- Whites of the Eyes (Icterus)
- Fatigue or Fussiness (Anemia)
- Swelling of the hands and feet (Dactylitis)
- Not enough red blood cells (Anemia)
- Blood gets trapped in the spleen (Acute splenic sequestration)

### Teens/Young Adults

- Leg ulcers
- Avascular necrosis (death of bone tissue because of the lack of blood supply)
- Acute chest syndrome
- Eye damage

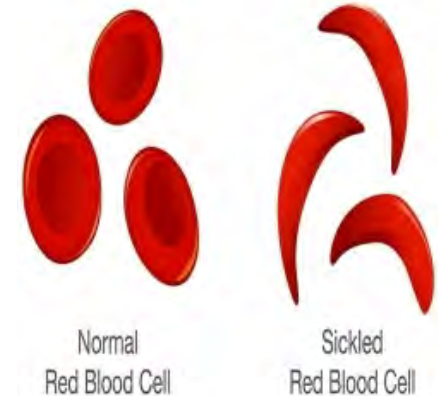
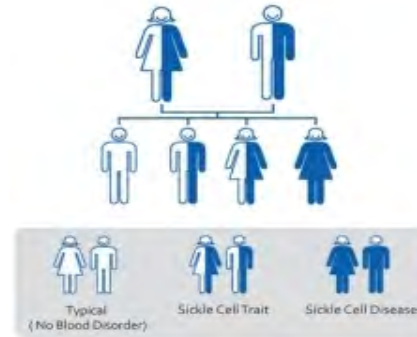
### Adults

- Pain Crisis caused by injury to the bones, muscle or internal organs

# Sickle Cell Disease

## Treatments

- Medications
- Blood Transfusions
- Stem Cell Transplant
- Hydroxyurea ( Taking this medication daily reduces pain and need for blood transfusion)
- L-glutamine oral powder (Endari: Helps reduce pain)
- Crizanlizumab (Adakveo: Reduces pain )
- Voxelotor (Oxbryta: Lowers risk of anemia and improves blood flow)



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