Gastrointestinal Disease in CGD

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Prevalence

- Studies report some GI disease in 27-44% of people with CGD
- Onset in x-linked disease by age 5
- Occurs more frequently in x-linked disease than autosomal recessive
Possible Symptoms

- Failure to thrive or falling off of the growth curve
- Weight loss
- Malaise and fatigue
- Abdominal pain
- Loose or frequent stools OR
- Bloating associated with infrequent stools/constipation
- Decreased appetite
- Fevers

SOMETIMES this is the presenting sign of CGD
Characteristics

- Strictures (narrowing) in the intestines
- Fistulas
- Obstruction secondary to granulomas
- Inflammatory process, not infectious
- Decreased protein or albumin, measured in the blood
- More lesions in lower colon (rectum and anus) but lesions are found throughout the GI tract from the mouth/esophagus on down
Diagnosis

- Endoscopy (upper and lower) is the Gold STANDARD for diagnosis
“Take home” for parents

- Be aware of the prevalence of GI disease associated with CGD
- Don’t discount subtle signs:
  - Fatigue
  - Changes in bowel habits
  - Changes in appetite
  - Abdominal pain
  - Weight loss
- Go with your parental instincts...if you think something is wrong, it probably is