Living with Chronic Granulomatous Disease (CGD)

What is Chronic Granulomatous Disease (CGD)?

Chronic Granulomatous Disease (CGD) is a genetic, primary immunodeficiency disorder in which white blood cells called phagocytes are unable to kill certain bacteria and fungi that enter the body.\(^1\) As a result, the immune system is compromised and patients are susceptible to infectious complications caused by various microbes.\(^2\)

CGD is characterized by severe, recurrent bacterial and fungal infections, and chronic inflammatory response, resulting in the formation of small areas of inflamed tissue (granulomas) and other inflammatory disorders.\(^3\)

Who Does CGD Impact?

Approximately one out of 200,000 people in the United States are born with CGD.\(^1\) The disorder may present at any time from infancy to late adulthood; however, the vast majority of affected individuals are diagnosed before the age of five.\(^3\)

People with CGD remain at risk for infection throughout their lives and often require hospitalization to treat infections.\(^2\) One study found that the most common cause of death for people with CGD was pneumonia and sepsis.\(^1\)

Despite the high health risks associated with the disease, the quality of life for people with CGD has improved dramatically in the past 50 years.\(^2\) The majority of children diagnosed with the condition can expect to live well into adulthood.\(^2\)

Early diagnosis is very important, so be sure to contact your doctor if you or your child experience any of these symptoms.\(^2\)

What are the Symptoms of CGD?\(^2\)

- Growth retardation in children
- Infections in the lungs, lymph nodes, liver, bones and skin
- Granuloma formations in the bladder and gastrointestinal track
- Colitis
- Abnormal wound healing

Even when there is not an infection, CGD can cause excessive inflammation that can contribute to diarrhea, bladder and kidney problems.\(^2\)

What Causes CGD?

CGD is an inherited genetic disorder that results from a flaw in a gene passed down from one or both parents. Since a parent can be a carrier,\(^4\) it’s very important to understand symptoms of CGD and the two ways it can be inherited:

1. The most common form of CGD – **X-linked CGD** – results from a mutation on the X chromosome,\(^2\) which is one of the two chromosomes that determine the sex of a child. It makes up 70 percent of the cases in the United States and affects almost only boys.\(^2\) For this form, the
mother is usually the carrier of the mutated X chromosome, and has a 50 percent chance of transmitting the gene to her children. While boys who inherit the mutation will be affected, females who inherit it will be carriers and will more than likely be asymptomatic.

2. CGD can also result from inheriting a mutation on an autosomal (non-sex determining) chromosome, of which there are 22. Known as **autosomal recessive CGD**, this form requires each parent to pass down the same mutated gene. Autosomal recessive CGD impacts both boys and girls equally.

### What Treatment Options are Available for CGD?

The goal of treatment for CGD is to prevent and reduce the frequency and severity of infections, prevent complications and halt an acute infection from becoming chronic and potentially causing irreversible damage.

Best practices for treatment for CGD can include the following:

- The aggressive use of appropriate antibiotics and antifungals is an important method to treat CGD infections. Use of the treatments prophylactically can help reduce infections as well.

In addition to treatment, CGD symptoms can be managed through careful lifestyle choices to avoid potentially harmful activities and environments:

- Activities like mulching and gardening should be avoided, as patients are exposed to decayed organic matter and can inhale fungal spores, which can cause infections.
- It’s important to avoid swimming in lakes, ponds or salt water, since dangerous organisms are frequently found there. A well-chlorinated pool is a safer option.

### References:


