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Chronic granulomatous disease

Chronic granulomatous disease (CGD) is an inherited disorder in which certain immune system cells do not function properly. CGD is characterized by repeated and severe infections.

Causes

In CGD, immune system cells called phagocytes are unable to kill some types of bacteria and fungi. This disorder leads to long-term (chronic) and repeated (recurrent) infections. The condition is often discovered very early in childhood. Milder forms may be diagnosed during the teenage years, or even in adulthood.

Risk factors include a family history of recurrent or chronic infections.

About half of CGD cases are passed down through families as a sex-linked recessive trait. This means that boys are more likely to get the disorder than girls. The variant gene is carried on the X chromosome. Boys have 1 X chromosome and 1 Y chromosome. If a boy has an X chromosome with the variant gene, he may inherit this condition. Girls have 2 X chromosomes. If a girl has 1 X chromosome with the variant gene, the other X chromosome may have a working gene to make up for it. A girl must inherit the variant X gene from each parent in order to have the disease.

Symptoms

CGD can cause many types of skin infections that are hard to treat, including:

- Blisters or sores on the face (impetigo)
- Eczema
- Growths filled with pus (abscesses)
- Pus-filled lumps in the skin (boils)

CGD can also cause:

- Persistent diarrhea
- Swollen lymph nodes in the neck
- Lung infections, such as pneumonia or lung abscess

Exams and Tests

Your health care provider will do an exam and may find:

- Liver swelling
- Spleen swelling
- Swollen lymph nodes

There may be signs of a bone infection, which may affect many bones.

Tests that may be done include:

- Bone scan
- Chest x-ray
- Complete blood count (CBC)
- Flow cytometry tests to help confirm the diagnosis
- Genetic testing to confirm the diagnosis
- Test of white blood cell function
- Tissue biopsy

Treatment

Antibiotics are used to treat infections caused by this condition. Chronic use of antibiotics may prevent infections. A medicine called interferon-gamma may also help reduce the number of severe infections. Surgery may be needed to treat some abscesses.

The only cure for CGD is a bone marrow or stem cell transplant.

Outlook (Prognosis)

Long-term antibiotic treatments may help reduce infections, but early death can occur from repeated lung infections.

Possible Complications

CGD may cause these complications:

- Bone damage and infections
- Chronic infections in the nose
- Pneumonia that keeps coming back and is hard to cure
- Lung damage
- Skin damage
- Swollen lymph nodes that stay swollen, occur often, or form abscesses that need surgery to drain them

When to Contact a Medical Professional

If you or your child has this condition and you suspect pneumonia or another infection, contact your provider right away.

Tell your provider if a lung, skin, or other infection does not respond to treatment.

Prevention

Genetic counseling is recommended if you are planning to have children and you have a family history of this disease. Advances in genetic screening and the increasing use of chorionic villus sampling (a test that may be done during a woman's 10th to 12th week of pregnancy) have made early detection of CGD possible.

Alternative Names

CGD; Fatal granulomatosis of childhood; Chronic granulomatous disease of childhood; Progressive septic granulomatosis; Phagocyte deficiency - chronic granulomatous disease

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