



What is chronic granulomatous disease?

Chronic granulomatous disease (CGD) is a very rare disease, which affects the way the immune system works. It is really not one single disease, but rather a group of related conditions. Some of the white blood cells do not work as well as they should, so children with CGD are prone to developing infections. CGD is a congenital disorder, which means that children are born with it. It affects about one in a million children. The word "granulomatous" relates to granulomas - scabs or scars which form in the tissues in response to infection.

Children with CGD may have one or more of the following:

- boils, abscesses (sores) in the neck or bottom
- pneumonia caused by certain bacteria and fungi
- fevers, abdominal pain and sometimes vomiting
- persistent rashes with skin scarring
- swollen lymph nodes that won't go down.

Why do children with CGD get infections?

White blood cells are produced in the bone marrow and then enter the blood to fight infections. There are two main types of white cells: the lymphocytes that fight viruses, and neutrophils (also called granulocytes or polymorphs) which fight bacteria and fungi.

Neutrophils "eat" bacteria and fungi and then use special proteins, called enzymes, to kill them. Neutrophils from children with CGD "eat" the bacteria and fungi but cannot kill them. This is

because children with CGD are born without one or more of the killing enzymes in their neutrophils.

How do you get CGD?

CGD is an inherited disease. You do not catch it, you are born with it. Most children with CGD (4 of every 5) are boys. The disease occurs because the affected gene, which tells white cells how to kill bugs, has a mistake in it (mutation) and doesn't work properly. The gene for one type of CGD (which affects only boys) is carried on the X-chromosome ("X" linked). The boys inherit their X-chromosomes from their mother. Mothers are usually the carrier, carrying the gene for CGD on one of their two X chromosomes, but never developing the disease themselves. If a mother carries the gene on one X chromosome, then half her sons on average will have CGD and half her daughters will be carriers. There is another type of CGD, which can affect either boys or girls. Affected children inherit one gene from each parent (autosomal recessive inheritance).

Is there a test for CGD?

- We can test for CGD using one of two special blood tests, either one called an NBT or a DMR test.
- Children with CGD will have abnormal neutrophils on either test.
- Mothers who are carriers of the X-linked type will usually have half their neutrophils abnormal. Because only a small number of neutrophils are needed to fight infection, carrier mothers usually have no signs or symptoms of CGD.
- The tests can check if a person has CGD, is a carrier of the X-linked type, or is normal.

Fact Sheet

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Can CGD be diagnosed during pregnancy?

Antenatal diagnosis in pregnancy is possible, usually by doing a test called a chorionic villus biopsy to look for the gene. This test can only be done if the exact mutation gene in the family has already been identified, otherwise an NBT or DMR test on a blood sample taken from the foetus can be done.

What is the outlook for affected children?

With modern care, children affected by CGD are living into adult life. The severity of the disease varies, so some children have lots of infections and others have very few. Doctors think that some children with CGD have a normal life expectancy.

Can CGD be treated?

The antibiotic cotrimoxazole (Bactrim, Septrin) has improved things enormously for children with CGD. Taken every day, it prevents many of the infections. Sometimes a second antibiotic must be used every day. Another option to reduce the number of severe infections is regular treatment with interferon-gamma. This drug is given three times a week by injection with a very small needle. Some children are given a special anti-fungal medicine to prevent fungal medicine.

Can it be cured?

The only possibility of cure is to perform a successful bone marrow transplant. Bone marrow transplant can be a dangerous procedure and may even be fatal, so it is not undertaken lightly and is not offered routinely. It will usually be discussed when the patient has a tissue matched sibling. Scientists have been interested in the possibility of using gene therapy (infecting CGD white cells with a virus carrying the normal gene for CGD) to cure CGD. Research programs are advancing rapidly, and it is hoped that in the future there may be more help or a cure for sufferers of the disease.

For more information

APIA (Australian Primary Immunodeficiency Association)

Telephone support and contact person:
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Phone: (02) 4942 1841
PO Box 121
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Immune Deficiency Foundation

PO Box 586
Columbia MD 21045
United States of America

CGD Support Group

Seafields, Shootersway Lane
Berkhamstead, Herts HP4 #Np England

Remember

- The antibiotic cotrimoxazole (Bactrim, Septrin) has improved lifestyle greatly for children with CGD.

This fact sheet is for education purposes only.
Please consult with your doctor or other health professional
to make sure this information is right for your child.

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hospital at Westmead

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