What Is Chronic Granulomatous Disease?

Chronic granulomatous disease (CGD) is a rare genetic disease that affects the immune system. In CGD, white blood cells can’t kill certain types of bacteria and fungi. As a result, people with CGD are more likely to get frequent infections. They are also more likely to have inflammation. Infections and inflammation can lead to chronic lumps called granulomas.

Known as a primary immune deficiency disease (PIDD), CGD affects white blood cells. White blood cells normally attack and destroy bacteria and fungi using chemicals that include bleach. In CGD, phagocytes—cells that eat up invaders—can’t make bleach, so they have trouble killing certain bacteria and fungi. When cells try to fight infections but can’t kill them, granulomas can form.

To prevent infections, people with CGD take medicines called antibiotics and antifungals. They may also take a medicine called interferon-gamma. Interferon-gamma is given as an injection and makes cells more able to kill bacteria and fungi. It also helps to reduce the number of severe infections in people with CGD. Even with treatment, people with CGD may develop abscesses—pus spots where infection has collected. Abscesses may need to be drained with surgery. People with lots of inflammation may take steroids, which are drugs that reduce inflammation. Some people with CGD have been treated successfully with bone marrow transplants. NIAID scientists are also working on ways to fix the genes that cause CGD.

*Terms in bold are defined in the glossary on page 4.*
Genetics and Inheritance

CGD is caused by problems in any of several genes.

Each person has 23 pairs of chromosomes. This includes one pair of sex chromosomes: males have one X and one Y chromosome, while females have two X chromosomes. The most common form of CGD is caused by changes in one of the genes on the X chromosome. If a male gets an X chromosome with a broken CGD gene, he will have CGD. But if a female gets one X chromosome with a broken CGD gene, she usually will be OK because she still has a working CGD gene on her other X chromosome. Although females who inherit a broken CGD gene on one X chromosome usually do not have CGD, they can still pass on the broken X chromosome to their sons, who then may have CGD.

In the United States, about two out of three CGD cases are caused by this X chromosome type of CGD. CGD can also be inherited through genes that are not on the X chromosome. In these cases, both males and females have an equal chance of having CGD.
Living with CGD can be hard for you and your family. It’s important for you and your family to talk about CGD and how the family is dealing with it so everyone can learn to cope with it as best they can. You and your family may find it helpful to meet and talk to other families affected by CGD. Patient organizations such as the Immune Deficiency Foundation (www.primaryimmune.org) or the CGD Society (cgdsociety.org) offer information on CGD and provide support for families affected by this disease.

Counseling can also help families cope with the challenges of CGD. Children learn who they can turn to for support and how to solve problems. Knowing the challenges of living with CGD can help everyone to grow stronger.
Abscess—A swollen area within body tissue, containing pus.

Bacteria—Microorganisms that can cause infection.

Cell—The basic unit of living organisms. Cells are made up of a nucleus (control center) and cellular organs wrapped in a membrane. Groups of cells with similar structures and functions form tissues.

Chromosomes—Physical structures in the cell nucleus that carry genes. Each human cell has 23 pairs of chromosomes.

Chronic—Going on for a long time or constantly recurring.

Colitis—When the large intestine is inflamed.

DNA (deoxyribonucleic acid)—Found in the cell nucleus, DNA carries genetic information.

Fungi—Yeasts and molds.

Gene—A unit of genetic material made up of DNA that controls something, like a protein or a process.

Granuloma—A lump of tissue produced in response to infection or inflammation. Granulomas form when the immune system is unable to eliminate infected tissue.

Immune system—A complex network of cells and proteins that defends the body against infection.

Inflammation—What happens when the body’s white blood cells and the substances they produce respond to infection or other things.

Inheritance—The passing of genetic traits.

Microorganisms—Microscopic living organisms, usually one-celled organisms. Microorganisms include bacteria and fungi.

Mutation—A change in the DNA sequence that is associated with disease.

Pneumonia—An infection in the lungs caused by bacteria, viruses, or fungi.

Primary immune deficiency disease (PIDD)—A genetic problem of the immune system.

Sex chromosomes—Females have two X chromosomes, while males have one X and one Y chromosome.