



Missing Mitochondrial Genes



Fact Sheet No.8

Lupus

Some people may know a little about lupus, which affects some carrier mothers. Our clinical nurse specialist provides a helpful explanation:

Mothers of males with CGD may be carriers of the X-linked form of CGD (XL-CGD). Carrier status can be determined by a simple blood test (NBT). In general carrier mothers are healthy, although in some cases may get recurrent mouth ulcers or even regular skin infections. If these are troublesome, prophylactic antibiotics could be tried. Occasionally carrier mothers may have symptoms associated with a condition called lupus.

What is Lupus?

There are two forms of lupus. One form, discoid lupus erythematosus (or DLE) is largely confined to the skin. The second form of this condition, systemic lupus erythematosus (SLE) is more generalized. Lupus is an autoimmune disease, caused by a fault in the body's immune system. Normally the immune system produces antibodies to help fight infection. In people who have autoimmune conditions antibodies are made which act against the body's own tissues, causing inflammation. These are called auto-antibodies. The common symptoms associated with lupus are skin rashes and joint pains but symptoms of the systemic condition may include inflammation of internal organs such as the lungs, heart, nervous system and kidneys.

How do I know if I have lupus?

Lupus is diagnosed using a wide variety of blood tests. These blood tests look for particular types of auto-antibodies. One of the most common tests looks for anti nuclear antibodies (ANA), and antibodies to double stranded DNA (which makes up genes and chromosomes). There are a number of CGD carrier mothers who have lupus-like symptoms. These are mainly skin rashes, which are very sensitive to the sun, joint pain and tiredness. We have called these symptoms lupus-like because most carrier mothers who have been tested for lupus show either negative blood tests or only weakly positive blood tests. At the Great Ormond Street clinic we asked 27 carriers of CGD whether they had any symptoms that were "lupus-like". 12 of them reported that they did have at least one symptom. These mothers then went on to have blood tests done. We found that most of them do not have positive blood tests for lupus despite having some symptoms.





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For many people lupus-like symptoms are no more than a nuisance, but some people who have troublesome symptoms may benefit from the treatments used in lupus, even if their lupus blood tests are negative. The systemic form of lupus can be a more unpredictable condition which should be carefully monitored by a Rheumatologist (often in a special 'lupus clinic'). If you are a carrier of CGD and think you may have lupus-like symptoms you should discuss these with your GP, get the screening tests done and consider assessment by a Rheumatologist. It is important that the GP (or rheumatologist) understands that there is a known link between lupus-like problems and being a carrier of CGD. The CGD nurses would be happy to discuss any concerns you may have or provide some information for your doctor.

What are lupus-like symptoms?

Lupus like skin rash often develops over parts of the body which are exposed to the sun, such as on the face, wrists/hands and chest. Some people find that they have problems with their hands in the cold with their skin becoming very white or blue coloured. This is very common problem, which is known as Raynaud's phenomenon joints. Many CGD carrier mothers have problems with their joints mostly involving pain and some swelling. Lupus-like symptoms are often associated with feeling very tired. This tends to come and go and people find that they have good days and bad days.

Treatment

People who have joint pain and skin rashes are often simply treated with creams for their skin rash (often containing some steroids) or anti-inflammatory medicines. Some people find that anti-malarial drugs (commonly Hydroxychloroquine) are also effective in treating the symptoms of lupus particularly the feelings of tiredness (these must be prescribed by a doctor specifically for this purpose). People who have more serious complications associated with systemic lupus may be treated with steroid tablets or other drugs which damp down the immune system (immunesuppressant medication). People on these medications need to be carefully monitored and have regular blood tests and review with the doctor.





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The skin rashes associated with Lupus are often very sensitive to the sun. So it is probably best to avoid sun bathing and wear a high factor sun cream, of Sun Protection Factor (SPF) 25 or greater (this may be available on prescription from your GP), when outside. People who have problems with Raynaud's phenomenon should keep warm and wear thick gloves and socks or tights. Tiredness tends to come and go, so it's probably best to take a break and try and get some rest on bad days and make the most of good days.

If you do have any 'lupus-like' symptoms it is best to get yourself checked out. So do speak with the CGD Nurses or your doctor.

Important Note: The information contained in this document is intended only as a guideline, not as a substitute for medical advice. Always consult your doctor if you or your child has any CGD symptoms or concerns.

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Manor Farm • Wimborne St. Giles • Dorset • BH21 5NL • ENGLAND

Phone/fax: 01725 517977 • e-mail: cgdresearchtrust@dial.pipex.com • www.cgd.org.uk

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