



Supporting families affected by chronic granulomatous disorder

CGD carriers and lupus-like symptoms
An information sheet for GPs

I am a carrier of X-linked Chronic Granulomatous Disorder, a genetic disorder which affects the immune system.

CGD carriers are unlikely to have the condition CGD but are known to be at risk from developing lupus-like symptoms.

It is recommended that CGD carriers presenting with aphthous ulceration, skin rashes, joint pains, fatigue and headaches are considered for referral to a rheumatologist and/or other relevant specialists for treatment of 'lupus-like symptoms', regardless of the results of a test for lupus. These lupus-like symptoms need to be taken seriously in CGD carriers even though lupus test results are likely to be negative. This is supported by the following evidence:

- **A Great Ormond Street Children's Hospital study**
'Cutaneous and other lupus-like symptoms in carriers of X-linked chronic granulomatous disease: incidence and autoimmune serology'. Clin Exp Immunol. 2007 Apr; 148(1):79-84. Cale CM, Morton L, Goldblatt D.

This concluded that symptoms of photosensitivity and skin rashes, joint pains, and fatigue and aphthous ulceration, are common in carriers of X-CGD. If these symptoms are significant, consideration should be given to referral to a rheumatologist or dermatologist and appropriate treatment should begin. Negative serology is probable, and should not influence diagnosis and treatment.

Visit www.cgdsociety.org/newlydiagnosed/carriermothers for other supporting evidence.

- **An informal CGD Society Carrier survey** in 2010 highlighted the symptoms experienced by X-CGD carriers. Of the 35 carriers included in the study, 85% had mouth ulcers, 88% were affected by skin problems, 68% experienced abnormal fatigue and 45% had recurrent headaches. Many people who responded had a combination of symptoms and some were severe and debilitating. The survey showed that symptoms tend to increase with age.

Visit the CGD Society website for more information about CGD carriers and lupus.

For more details, please contact:

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You may find it useful to scan this sheet into your patient notes.