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Majeed Syndrome

Version of 2016

2. DIAGNOSIS AND THERAPY

2.1 How is it diagnosed?

The disease should be suspected on the basis of the clinical presentation. The definitive diagnosis must be confirmed by genetic analysis. The diagnosis is confirmed if the patient carries 2 mutations, one from each parent. Genetic analysis may not be available in every tertiary care centre.

2.2 What is the importance of tests?

Blood tests such as erythrocyte sedimentation rate (ESR), CRP, whole blood count and fibrinogen are important during disease activity to assess the extent of inflammation and anaemia.
These tests are periodically repeated to evaluate if the results are back to or near normal. A small amount of blood is also needed for the genetic analysis.

2.3 Can it be treated or cured?

Majeed syndrome can be treated (see below) but not cured since it is a genetic disease.

2.4 What are the treatments?

There is no standardised therapeutic regimen for Majeed syndrome. CRMO is usually treated, as a first line, with non-steroidal anti-inflammatory drugs (NSAIDs). Physical therapy is important to avoid

disuse atrophy of muscles and contractures. If CRMO does not respond to NSAIDs, corticosteroids can be used to control CRMO and skin manifestations; however, the complications of long-term use of corticosteroids limit their use in children. Recently, good response to anti-IL1 drugs has been described in 2 related children. CDA is treated with red blood cell transfusion if indicated.

2.5 What are the side effects of drug therapy?

Corticosteroids are associated with possible side effects such as weight gain, swelling of the face and mood swings. If the steroids are prescribed for a prolonged period, they may cause suppression of growth, osteoporosis, high blood pressure and diabetes.
The most troublesome side effect of anakinra is the painful reaction at the site of injection, comparable to an insect sting. Especially in the first weeks of treatment, these may be quite painful. Infections have been observed among patients treated with anakinra or canakinumab for diseases other than Majeed Syndrome.

2.6 How long should treatment last?

Treatment is life-long.

2.7 What about unconventional or complementary therapies?

There are no known complementary therapies for this disease.

2.8 What kind of periodic check-ups are necessary?

Children should be seen regularly (at least 3 times yearly) by a paediatric rheumatologist to monitor the control of the disease and adjust the medical treatment. Periodic complete blood count (CBC) and acute phase reactants should be carried out to determine if red blood cell transfusion is necessary and to evaluate the control of inflammation.

2.9 How long will the disease last?

This disease is life-long. However, the activity of the disease may

fluctuate over time.

2.10 What is the long-term prognosis (predicted outcome and course) of the disease?

The long-term prognosis depends on the severity of clinical manifestations, particularly on the severity of dyserythropoietic anaemia and disease complications. If left untreated, the quality of life is poor as a result of recurrent pain, chronic anaemia and possible complications including contractures and disuse atrophy of the muscles.

2.11 Is it possible to recover completely?

No, because it is a genetic disease.