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Chronic Atypical Neutrophilic Dermatosis with Lipodystrophy and Elevated Temperature (CANDLE)

Version of 2016

2. DIAGNOSIS AND TREATMENT

2.1 How is it diagnosed?

First there has to be a suspicion of CANDLE based on the disease features. CANDLE can only be proven by genetic analysis. The diagnosis of CANDLE 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 carried out during disease activity to evaluate the extent of inflammation and anaemia; tests of liver enzymes are performed to assess liver involvement.
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?

CANDLE cannot be cured since it is a genetic disease.

2.4 What are the treatments?

There is no effective therapeutic regimen for CANDLE syndrome. High doses of steroids (1-2 mg/kg/day) have been shown to improve some symptoms including skin eruptions, fever and joint pain but once tapered, these manifestations often return. Tumour necrosis factor alpha (TNF-alpha) inhibitors have provided temporary improvement in some patients but gave rise to flares in others. The immunosuppressive drug, tocilizumab, has shown minimal efficacy. Experimental studies with the use of JAK-kinase inhibitors (tofacitinib) are ongoing.

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 can cause suppression of growth, osteoporosis, high blood pressure and diabetes.
TNF-α inhibitors are recent drugs; they can be associated with an increased risk of infection, activation of tuberculosis and possible development of brain or other immune diseases. A potential risk of development of malignancies has been discussed; at present, there are no statistical data proving an increased risk of malignancies with these drugs.

2.6 How long should treatment last?

Treatment is life-long.

2.7 What about unconventional or complementary therapies?

There is no evidence concerning this type of therapy for CANDLE Syndrome.

2.8 What kind of periodic check-ups are necessary?

Children should be seen regularly (at least 3 times yearly) by their paediatric rheumatologist to monitor the control of the disease and adjust the medical treatment. Children being treated should have blood and urine tests at least twice yearly.

2.9 How long will the disease last?

CANDLE is a life-long disease. 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?

Life expectancy can be reduced, with death often the result of multi-organ inflammation. Quality of life is often poor as patients suffer from reduced activity, fever, pain and repeated episodes of severe inflammation.

2.11 Is it possible to recover completely?

No, because it is a genetic disease.