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NLRP-12 Related Recurrent Fever

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2. DIAGNOSIS AND TREATMENT

2.1 How is it diagnosed?

A medical expert will suspect this disease on the basis of clinical symptoms identified during a physical examination and from taking a family medical history.
Several blood analyses are useful to detect inflammation during the attacks. The diagnosis is confirmed only by genetic analysis providing evidence of mutations. Differential diagnoses are other conditions presenting with recurrent fever, especially cryopyrin-associated periodic syndromes.

2.2 What is the importance of tests?

As above, laboratory tests are important in diagnosing NLRP-12-related recurrent fever. Tests such as CRP, serum Amyloid A protein (SAA) and whole blood count are important during an attack to assess the extent of inflammation.
These tests are repeated after the child becomes symptom-free to observe 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?

NLRP-12-related recurrent fever cannot be cured. There is no effective preventive treatment for attacks. Treating the symptoms can reduce inflammation and pain. Some new drugs to control inflammatory symptoms are currently under investigation.

2.4 What are the treatments?

The treatments for NLRP-12-related recurrent fever include non-steroidal anti-inflammatory drugs such as indomethacin, corticosteroids such as prednisolone and possibly biologic agents such as anakinra. None of these drugs appears to be uniformly effective, although all of them appear to help in some patients. Proof of their efficacy and safety in NLRP-12-related recurrent fever is still lacking.

2.5 What are the side effects of drug therapy?

Side effects depend on the drug that is used. NSAIDs may cause headaches, stomach ulcers and kidney damage, corticosteroids and biologic agents may increase susceptibility to infections. In addition, corticosteroids may cause a wide variety of other side effects.

2.6 How long should treatment last?

There are no specific data to support life-long therapy. Given the normal tendency for improvement as patients grow up, it is probably wise to attempt drug withdrawal in patients whose disease appears to be quiescent.

2.7 What about unconventional or complementary therapies?

There are no published reports of effective complementary remedies.

2.8 What kind of periodic check-ups are necessary?

Children affected with NLRP-12-related recurrent fever should have blood and urine tests at least twice a year.

2.9 How long will the disease last?

The disease is life-long, although symptoms may get milder with age.

2.10 What is the long-term prognosis (predicted outcome and

course) of the disease?

NLRP-12-related recurrent fever is a life-long disease, although symptoms may get milder with age. As the disease is very rare, the exact long-term prognosis is still unknown.