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Mevalonate Kinase Deficiency (MKD), or Hyper IGD Syndrome

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2. DIAGNOSIS AND TREATMENT

2.1 How is it diagnosed?

The diagnosis is based on chemical studies and genetic analysis. Chemically, abnormally high mevalonic acid can be detected in urine. Specialised laboratories can also measure the activity of mevalonate kinase enzyme in the blood or skin cells. Genetic analysis is performed on DNA of the patient, in which the mutations to the MVK genes can be identified.
Measurement of serum IgD concentration is no longer considered a diagnostic test for mevalonate kinase deficiency.

2.2 What is the importance of tests?

As mentioned above, laboratory tests are important in diagnosing mevalonate kinase deficiency.
Tests such as erythrocyte sedimentation rate (ESR), CRP, serum Amyloid-A-protein (SAA), whole blood count and fibrinogen 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 sample of urine is also tested for the presence of protein and red blood cells. There may be temporary changes during attacks. Patients with amyloidosis will have persistent levels of protein in urine tests.

2.3 Can it be treated or cured?

The disease cannot be cured, nor is there a proven effective treatment to control disease activity.

2.4 What are the treatments?

The treatments for mevalonate kinase deficiency include non-steroidal anti-inflammatory drugs such as indomethacin, corticosteroids such as prednisolone and biologic agents, such as etanercept or anakinra. None of these drugs appears to be uniformly effective but all of them appear to help in some patients. Proof of their efficacy and safety in mevalonate kinase deficiency 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 increase susceptibility to infections. In addition, corticosteroids may cause a wide variety of other side effects.

2.6 How long should treatment last?

There is no data to support life-long therapy. Given the normal tendency for improvement as patients grow older, 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 being treated should have blood and urine tests at least twice yearly.

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?

Mevalonate kinase deficiency is a life-long disease, although symptoms may get milder with age. Very rarely, patients develop organ damage, especially to the kidneys, due to amyloidosis. Very severely affected patients may develop mental impairment and night-blindness.

2.11 Is it possible to recover completely?

No, because it is a genetic disease.