PATIENT FACT SHEET

Hyperimmunoglobulin-D Syndrome (Juvenile)

CONDITION DESCRIPTION

Mevalonate kinase deficiency (MKD), also known as hyperimmunoglobulin-D syndrome, is a very rare, genetic disease in children. It is caused by an abnormal gene, MVK, and is mostly found in children in Western Europe. MKD affects both boys and girls, and symptoms usually appear in early childhood.

Mevalonate kinase is involved in cholesterol production, so MKD is a metabolic disease. Parents may be carriers of the gene and not have any illness. Patients make high amounts of immunoglobulins, especially D, which the immune system normally uses to fight infections.

SIGNS/ SYMPTOMS

Fevers are the main symptom of MKD. They may last for 3-7 days and recur every 2-12 weeks. Episodes begin suddenly, usually with chills. Children often have headaches, abdominal pain, appetite loss, flu-like symptoms, nausea, vomiting and diarrhea. Skin rashes all over the body, including painful mouth sores, may occur. Children may have joint pain and swelling, or muscle aches. Swollen lymph nodes are a striking feature.

Diagnosis of MKD must be made by a physician with expertise in this type of disease. During an episode, blood tests may show high levels of inflammation. Most children, but not all, will test for high levels of immunoglobulin-D. Younger children may not test high for this protein.

During episodes, urine tests will show high levels of mevalonic acid. A genetic test to show the abnormal MVK gene will confirm the diagnosis.

COMMON TREATMENTS

There is no cure for mevalonate kinase disease/ hyperimmunoglobulin-D syndrome, but treatments may help control inflammation and symptoms during episodes. Children may take nonsteroidal anti-inflammatory drugs (NSAIDs) like ibuprofen (Advil, Motrin) or naproxen (Aleve, Naprosyn), or corticosteroids during an episode.

Biologic medications that block proteins, like tumor necrosis factor or interleukin-1, have been found to be effective treatments to reduce the frequency of attacks. Anakinra (Kineret) may be an "on-demand" treatment used as an episode starts. Canakinumab (Ilaris) and etanercept (Enbrel) also may prevent attacks in patients with more severe disease.

CARE/ MANAGEMENT TIPS

MKD episodes usually get milder and less frequent as children grow up, and resolve by adulthood. However, children may miss many days of school due to frequent episodes. It is important to stress to other children and classmates that the fevers related to MKD are not contagious.

Patients rarely may develop a complication of inflammation called amyloidosis. This is a build-up of amyloid proteins which then deposit into organs such as the kidneys, but possibly the heart, skin or intestines as well. Amyloidosis may cause loss of organ function, especially kidney function. Regular medical check-ups to monitor for amyloidosis and other potential problems are important.

Updated March 2017 by Jennifer Murphy, MD, and reviewed by the American College of Rheumatology Committee on Communications and Marketing.