Neonatal-Onset Multisystem Inflammatory Disease (NOMID): Fact Sheet

TOPICS:

What It Is

Neonatal-Onset Multisystem Inflammatory Disease (NOMID) (also called Chronic Infantile Neurologic Cutaneous Articular, or CINCA, Syndrome) is a subtype of Cryopyrin-Associated Periodic Syndromes (CAPS). Symptoms generally appear shortly after birth. NOMID is the most severe form of CAPS.[1]

Causes and Symptoms

NOMID is generally caused by mutations in a gene called *NLRP3* (nucleotide-binding domain, leucine-rich family [NLR] pyrin domain containing 3). *NLRP3* mutations cause increased activity of cryopyrin, a protein that regulates inflammation. Increased cryopyrin activity causes overproduction of a protein called interleukin-1 beta (IL-1β).[1,2]

Overproduction of IL-1β, in turn, causes recurrent symptoms of inflammation such as[2]

- Rash
- Fever/chills
- Joint pain
- Eye redness/pain
- Fatigue

NOMID Characteristics

Central nervous system symptoms include chronic meningitis, mental impairment, seizures, and sensory organ dysfunction, which results in vision and hearing loss. Joint inflammation leading to joint and bone deformities varies in severity and typically begins in the knees.[1] Enlargement of the kneecaps may also occur. Other symptoms include increased levels of white blood cells, enlargement of the liver and spleen, and abnormal facial features.[3]

Symptoms are similar to (and should not be confused with) juvenile idiopathic arthritis (JIA). High, recurrent fevers, as well as joint pain, deforming joint disease, and rash, are symptoms of both NOMID and JIA.[1,4] However, NOMID is differentiated by the onset of skin disease at birth and a persistent rash.[1]

Living with NOMID

Patients may have frequent, almost daily flares that cause great discomfort and may require medical assistance. Some patients are unable to walk or bear weight on their legs because of joint damage or pain. Most children with NOMID have cognitive and mental deficits, as well as learning disabilities and vision and hearing loss.[3]

Diagnosis and Treatment

Diagnosis of NOMID is determined through an evaluation of a patient's symptoms and medical history. Confirmation of the diagnosis is sometimes achieved through genetic testing and the identification of *NLRP3* mutations.[3] Not all patients, however, have a detectable genetic mutation, making accurate symptom evaluation critical.[1]

There are no therapies indicated specifically for the treatment of NOMID.