

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

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What It Is

Neonatal-Onset Multisystem Inflammatory Disease (NOMID), also known as Chronic Infantile Neurologic Cutaneous Articular (CINCA) syndrome, is a rare, congenital, systemic, inflammatory condition distinguished by recurrent rash, fever/chills, joint pain, fatigue, eye pain/redness, and central nervous system (CNS) disease. Symptoms of NOMID generally appear shortly after birth. NOMID is the most severe form of the Cryopyrin-Associated Periodic Syndromes (CAPS) oftentimes caused by mutations in the CIAS1/NLRP-3 gene.

Causes and Symptoms

Many patients with NOMID have alterations in a gene identified as the Cold-induced Auto-inflammatory Syndrome 1 (CIAS1) gene, more recently named the Nod-Like Receptor Protein-3 (NLRP-3) gene. The mutation in the CIAS1/NLRP-3 gene causes increased activity of cryopyrin, a protein that regulates inflammation in the body. The increased activity of cryopyrin results in an overproduction of a protein known as interleukin-1 β (IL-1 β), which leads to symptoms of inflammation such as fever and joint pain.

NOMID Characteristics

Symptoms of NOMID generally include recurrent rash, fever/chills, joint pain, fatigue, and eye pain/redness. These symptoms occur in all patients within the first six weeks of life and persist throughout their lives. CNS symptoms include chronic meningitis, mental retardation, seizures, and sensory organ dysfunction, which results in vision and hearing loss. Joint inflammation leading to joint and bone deformities ranges in severity and typically begins in the knees. Enlargement of the knee-cap is also characteristic of NOMID.

Other symptoms include stunted growth, enlargement of the liver and spleen, an abnormal increase in the number of white blood cells, an elevation in levels of the protein amyloid A and C-reactive protein in the blood, and an increase in the erythrocyte sedimentation rate, a blood test used to detect or grade inflammation. In addition, abnormal facial features can sometimes be seen.

NOMID shares symptoms, and should not be confused, with [juvenile idiopathic arthritis \(JIA\)](#). High recurrent fevers, joint pain, deforming joint disease, and rash are symptoms of both NOMID and JIA. However, NOMID is differentiated by the onset of skin disease at birth and a persistent rash. In addition, many patients with NOMID have nonspecific joint pain and enlargement of the knee-cap, while patients with JIA present with inflamed synovial joints, such as the shoulder or knee, increased production of fluid in the synovial joints and warm, swollen, stiff joints.

Incidence and Prevalence

NOMID is considered a very rare disease. Since NOMID is a newly discovered condition, the actual incidence and prevalence of the disease is difficult to determine.

Living with NOMID

NOMID patients suffer from frequent, almost daily flare-up episodes which cause great discomfort, can be very debilitating, and may require medical assistance during the episodes. Some patients are unable to walk, or bear weight on their legs, due to joint damage, and/or pain. The majority of children with NOMID have cognitive and mental deficits, and/or learning disabilities, as well as vision and hearing loss. There is a 20 percent mortality rate before adulthood associated with NOMID.

Diagnosis

Diagnosis of NOMID is determined through an evaluation of a patient's symptoms and medical history. Confirmation of the diagnosis is oftentimes, but not always, achieved through genetic testing.