

Section: CLINICAL CASE REPORT

Abstract: CCR-07

CINCA/NOMID syndrome: early diagnosis and anti-interleukin-1 treatment

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INTRODUCTION: Chronic infantile neurologic, cutaneous, and articular (CINCA) syndrome, also known as neonatal-onset multisystem inflammatory disease (NOMID), is characterized by chronic systemic inflammation caused by dysregulation of the innate immune system and is included within cryopyrin-associated periodic syndromes (CAPS). The autoinflammatory abnormality in this disease results from a mutation in NLRP3, a molecule that regulates the activation and secretion of interleukin-1 beta, which is central to regulation of the inflammatory response. **CASE REPORT:** A five-month-old boy was the first child of healthy, nonconsanguineous parents. Soon after birth, he developed multiple scaly erythematous lesions throughout the body, followed by an abdominal abscess, hepatitis, meningitis, and pyogenic arthritis, with no etiologic agents isolated. After extensive clinical investigation, inflammatory markers remained elevated (erythrocyte sedimentation rate, C-reactive protein, and serum amyloid A), and leukocytosis with a left shift was present in peripheral blood. An autoinflammatory disease was suspected, and a customized multigene panel was requested. A probably pathogenic heterozygous p.Gly309Val variant in NLRP3 was identified, confirming CINCA syndrome. The patient remained hospitalized for 51 days until clinical stabilization. After discharge, prednisolone 1.5 mg/kg/day and canakinumab (anti-interleukin-1) every 15 days were initiated, with a good clinical response. After the second dose of the biologic agent, the skin lesions resolved completely, weight gain and neuropsychomotor development improved, joint edema decreased, and no hearing loss was detected. **DISCUSSION:** CINCA syndrome is part of the cryopyrin-associated periodic syndromes. It is classified as an innate immune defect with excessive inflammasome activation, resulting in IL-1 secretion and dysregulated activation of pyroptosis. This activates inflammatory pathways, notably neutrophil recruitment and associated febrile episodes. Clinical organ damage affects the skin (neutrophilic urticaria), central nervous system (meningitis and deafness), and joints (arthritis). Treatment consists of corticosteroids, which nonspecifically inhibit inflammatory cytokines, or specific IL-1 blockade. Available anti-IL-1 agents include canakinumab, anakinra, and rilonacept.

KEYWORDS: CINCA-NOMID; Autoinflammatory disease; Innate immunity.