Neonatal Onset Multisystem Inflammatory Disease Presenting as an Urticarial Rash in a Newborn

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Introduction

Neonatal Onset Multisystem Inflammatory Disease (NOMID) is a rare periodic fever syndrome associated with significant morbidity that increases with diagnosis and treatment delays. NOMID is the most severe and rare form of Cryopyrin Associated Periodic Syndromes (CAPS), with around 100 cases described in the literature. NOMID is caused by gain-of-function mutations in NLRP3 gene which activates cryopyrin inflammasome, leading to inappropriately increased innate immune response and multisystem dysfunction. Clinically, this leads to sensorineural hearing loss, vision loss, intellectual disability and seizures secondary to sterile meningitis and increased intracranial pressure, arthropathy and renal failure. This case describes the youngest patient to have been started on Anakinra for NOMID.

Case Description

A 6-day-old male presenting for evaluation of a waxing and waning urticarial rash, which started at 9 hours of life. He had no fevers and an otherwise negative review of systems. On exam, the patient had blanching, raised, small erythematous papules on the lower extremities and abdomen. At first, the pediatric team suspected erythema toxicum, but further search of possible etiologies raised concerns for neonatal lupus or autoinflammatory diseases. Rheumatology was consulted, laboratory tests were unremarkable with the exception of thrombocytosis and elevated inflammatory markers. Genetic testing for CAPS was positive for heterozygous D303G mutation in the NLRP3 gene. The patient was diagnosed with NOMID. Treatment with daily subcutaneous Anakinra (IL-1 receptor antagonist) was initiated at 11-weeks of age with resolution of the rash within hours after the first injection. Inflammatory markers subsequently normalized. His development is appropriate, and he remains otherwise asymptomatic.

Discussion

This case illustrates the importance of prompt and accurate diagnosis of NOMID and early initiation of IL-1 targeted therapy. This case is unique in that diagnosis was made early, allowing rapid initiation of therapy that may prevent significant multisystem dysfunction including sensorineural hearing loss, uveitis and vision loss, progressive chronic meningitis and increased intracranial pressure leading to developmental delay, intellectual disability, and seizures, degenerative arthropathy with joint contractures, and AA amyloidosis leading to renal failure. The primary pediatrician plays an essential role in identifying atypical rashes that may be indicative of multisystemic and autoinflammatory diseases.

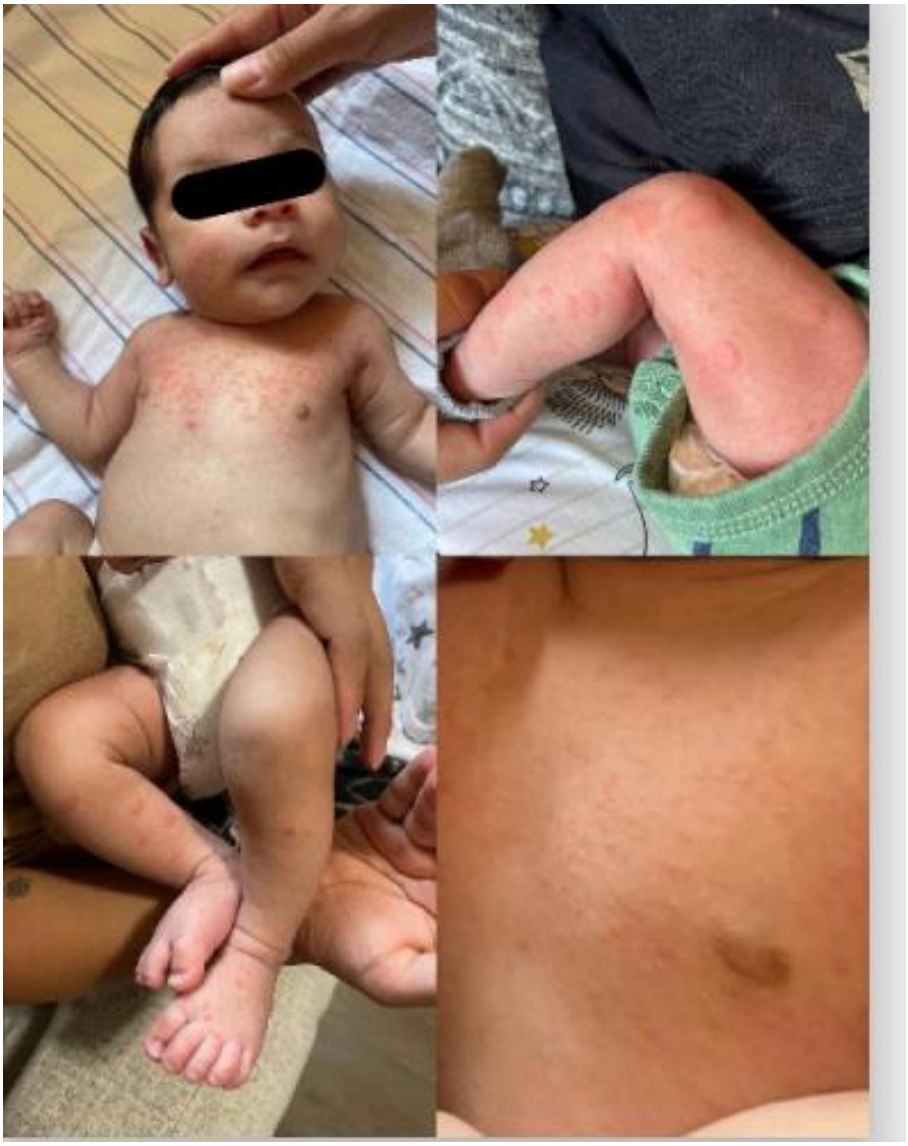

Figure 1: Rash from Neonatal Onset Multisystem Inflammatory Disease (NOMID) on newborn patient involving chest, face, and lower extremities