

# Bilateral Serous Retinal Detachment from Neonatal-Onset Multisystemic Inflammatory Disorder

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## ABSTRACT

**PURPOSE:** Neonatal-onset Multisystem Inflammatory Disorder (NOMID) is a systemic syndrome characterized by rash, large joint osteoarthropathies and chronic meningitis. Ocular manifestations include optic disc edema, corneal opacities and uveitis. We report the novel finding of serous retinal detachments (RD) in NOMID.

**METHODS:** Case report.

**RESULTS:** An eight-month-old girl was referred to the ED for work-up of optic disc edema. Physical exam revealed flat fontanelles; macrocephaly and frontal bossing; diffuse urticarial rash; and swollen joints; WBC count and inflammatory markers were elevated. Ophthalmology exam revealed decreased visual acuity, optic disc edema and bilateral serous RD. MRI revealed bilateral enhancement of the ocular choroid and enlargement of the third and fourth ventricles secondary to aqueductal webbing. After infectious testing returned negative, the patient was treated with anakinra, an interleukin-1 receptor antagonist. Three months later, the serous RDs resolved.

**CONCLUSION:** Physicians should consider NOMID in infants presenting with diffuse rash, bilateral disc edema and serous retinal detachments.

**KEYWORDS:** inflammatory disease; neonatal-onset multi-inflammatory disorder; ophthalmic manifestations; pediatric uveitis; serous retinal detachment

## INTRODUCTION

We present a novel finding of bilateral serous retinal detachment in an 8-month-old girl with Neonatal-onset Multisystem Inflammatory Disorder (NOMID). NOMID is a newly recognized autoimmune disorder, characterized by diffuse rash, large joint osteoarthropathies and chronic meningitis. Previously reported ophthalmic manifestations include optic disc edema, optic disc atrophy and uveitis. Knowledge of this syndrome is crucial because systemic and ocular inflammation respond well to treatment with anakinra, a systemic interleukin-1 antagonist.

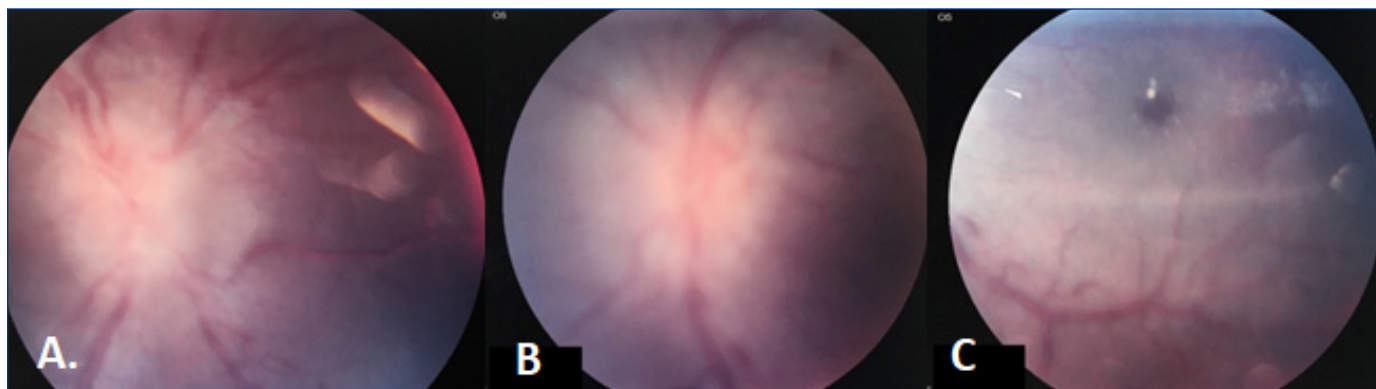
## CASE REPORT

An 8-month-old Hispanic girl presented to the emergency department (ED) after her pediatric ophthalmologist noted bilateral optic disc edema. The patient was born at 32 weeks gestation via vaginal delivery to a gravida 4 para 4 obese mother with gestational diabetes. At birth she had abnormal facies, diffuse rash, bilateral hearing loss and a placental appearance concerning for candidal infection. She was treated with systemic antifungal medication and admitted to the neonatal intensive care unit. A dilated retina examination performed by ophthalmology was normal at that time.

**Figure 1.** External photos depicting urticarial-like rash present over trunk and extremities (top) and facial rash with saddle nose deformity and frontal bossing (bottom).



**Figure 2.** Ocular fundus photos showing optic disc edema in the right (A) and left (B) eyes as well as the gray-colored retina (C).

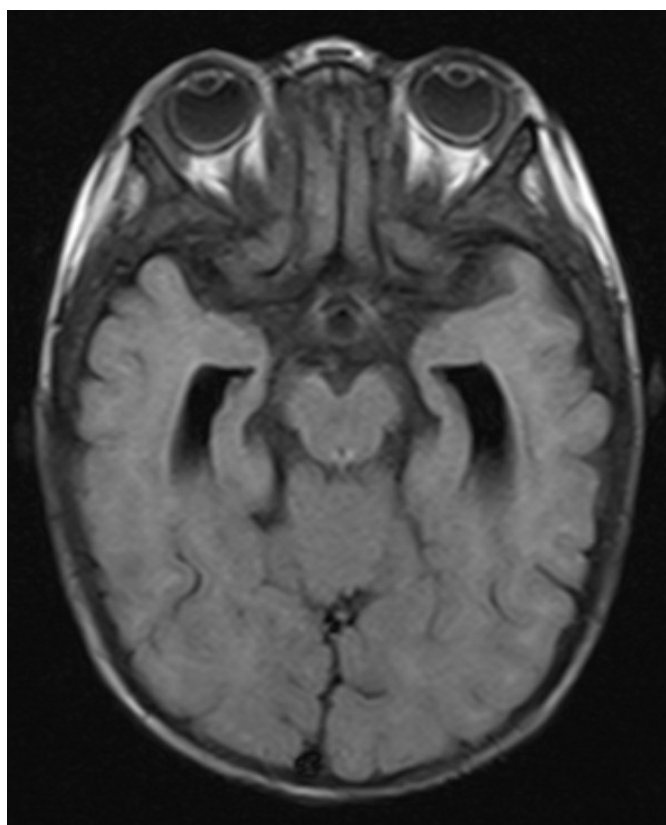


Her medical history included small vessel calcific thrombus of the left kidney and autoamputation of the left toe after peripheral IV infiltration with thrombus. Infectious, hypercoagulability and genetic microarray studies were negative.

Physical exam in the emergency room revealed flat fontanelles, macrocephaly with frontal bossing and saddle back nose deformity, urticarial rash on the face, trunk and extremities, and swollen joints (**Figure 1**). There was no fever. Her labs included CRP 140 mg/L (N=0.00-10 mg/L), ESR >140 mm/h (N=0.00-20 mm/h), WBC 27,000 cells/mcL (N=5,000-15,000 cells/mcL) and hemoglobin 9.3 g/dL (N=10.5-13.5 g/dL). Eye examination showed intermittent blinking to light and poor fixation and follow, which is lower visual acuity than expected for her age. Anterior segment examination with a portable slit-lamp was normal. A red reflex was blunted but visible. Dilated retinal examination revealed optic disc edema, tortuous vessels, and a gray colored retina bilaterally (**Figure 2**). Ocular ultrasonography showed a shallow inferior serous retinal detachment in both eyes. Magnetic resonance imaging with venography revealed increased enhancement of the ocular choroid bilaterally (**Figure 3**) and enlargement of the third and fourth ventricles secondary to aqueductal webbing. These findings were consistent with chronic hydrocephalus without increased intracranial pressure. Neurosurgery consultation recommended refraining from lumbar puncture or other surgical intervention. Tuberculosis and toxoplasmosis screening were negative. Anakinra, an interleukin-1 receptor antagonist, was initiated for presumed NOMID.

At four weeks after discharge, her ability to fixate had improved. Retinal examination revealed unchanged optic disc edema, resolution of the serous retinal detachments, and residual choroidal thickening on ocular ultrasound. At 10 weeks, ophthalmic examination revealed anterior vitreous cells with a portable slit-lamp. In consultation with rheumatology, her anakinra dose was increased with interval normalization of CRP level as well as complete resolution of fevers and rash. Her last visual acuity, three months after initial presentation, was central, steady and maintained with good fixation and follow in both eyes.

**Figure 3.** Bilateral ocular choroidal enhancement on axial FLAIR views of MRI. Lateral ventricular dilation is also noted.



## DISCUSSION

Neonatal-onset multisystem inflammatory disease (NOMID) is a syndrome characterized by persistent urticarial-like rash, large joint osteoarthropathies and chronic meningitis.<sup>1,2</sup> Since its recognition as a syndrome, apart from juvenile idiopathic arthritis, additional sequelae have been identified in other systems including the eyes. The most common ocular manifestation is optic disc edema eventually resulting in atrophy likely secondary to either neutrophilic infiltration<sup>3</sup> or chronic papillitis.<sup>4</sup> Additional ocular findings include perilimbal injection, corneal opacities, uveitis and vascular

sheathing.<sup>3,5</sup> Systemic inflammation is secondary to a mutation of the NLRP3/CIAS1 gene resulting in an upregulation of IL-1 production,<sup>6</sup> which responds to treatment with anakinra, a systemic IL-1 receptor antagonist.<sup>7</sup> We postulate that the etiology of this patient's serous retinal detachments is multifactorial. It is well documented that optic disc edema due to anterior ischemic optic neuropathy,<sup>8</sup> papilledema<sup>9</sup> and diabetic papillitis<sup>10</sup> disrupts glial tissue at the disc resulting in retinal separation and accumulation of subretinal fluid. However, this patient's serous retinal detachments resolved with immunosuppression despite persistent optic disc edema suggesting that breakdown of the blood-retina barrier in the setting of chronic inflammation also may have been a contributing factor.

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## Presented

Poster, American Society of Retina Specialists, Annual Meeting, Boston MA. August 2017.

## Acknowledgments

The authors thank the staff of Dr. Lory Snady-McCoy for their photography and Drs. Paul Greenberg and Magdalena Krzystolik for their editorial assistance. Thanks also to Dr. Altin Pani for his inpatient retinal consultation.

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