



Optic Disc Drusen in a Child: A Challenging Differential Diagnosis - A Case Report

Salvatore M Carnazzo^{1*}, Desirèe Balconara¹, Francesco Caruso² and Piero Pavone³

¹Postgraduate Training Program in Pediatrics, Department of Clinical and Experimental Medicine, University Hospital "Policlinico-San Marco of Catania, Italy

²Postgraduate Training Program in Hygiene and Preventive Medicine Department of Biomedical Sciences, University of Catania, Italy

³Acute End Emergency Pediatric Unit, Auo Policlinico 'Rodolico-san Marco' University of Catania, Italy

***Corresponding Author:** Salvatore M Carnazzo, Postgraduate Training Program in Pediatrics, Department of Clinical and Experimental Medicine, University Hospital "Policlinico-San Marco of Catania, Italy.

Received: March 22, 2023

Published: May 23, 2023

© All rights are reserved by **Salvatore M Carnazzo, et al.**

Abstract

Optic disc drusen are calcified deposits located at the anterior optic nerve head that can cause symptoms such as amaurosis or visual defects if they are voluminous. This article discusses the pathophysiology, diagnosis, prevalence, and associated complications of optic disc drusen, highlighting the need for differential diagnosis between optic disc drusen and papilledema, which can be challenging, leading to unnecessary investigations.

The article presents a case of a 5-year-old boy who presented with symptoms of headache, pain with eye movements, vomiting, and vision loss, which was ultimately diagnosed with optic disc drusen. The article emphasizes that a correct diagnosis of optic disc drusen cannot be made based solely on clinical history and funduscopy and discusses the effectiveness of different diagnostic methods, including ultrasound.

This case report emphasizes the importance of differential diagnosis between optic disc drusen and papilledema and highlights the need for clinicians to be aware of the challenges in diagnosing optic disc drusen. A correct diagnosis of optic disc drusen is crucial in avoiding unnecessary and invasive investigations. The article concludes by discussing the clinical relevance of optic disc drusen and their potential to cause symptoms and be recognized in several pathologies.

Keywords: Optic Disc Drusen; Optical Coherence Tomography; Papilledema; Misdiagnosis; Visual Evoked Potentials; Pediatric; Fundus Oculi Examination

Introduction

After ethical and family approval we report a forty-eight year old white male with no medical history, underwent surgery for acute hemorrhoids. Shortly after, the patient reported fatigue, arthralgia and mild fever. No local surgical complications were found. The following day, the patient was admitted to intensive care unit in Mahdia teaching Hospital (Tunisia) for multiple organ failure. Physical examination showed high persistent fever (40°C) despite antipyretics, severe hypotension, confusion, anuria, cardiac arrhythmia, rash on the chest and abdomen, mucositis, red dry cracked lips and swollen tongue, conjunctivitis and skin ulcerations on genital area (Images were obtained with patient consent and are shown in the appendix). Blood tests revealed an inflammatory state: C-reactive protein (CRP) level was 266 mg/L, anemia

(Hb: 8.1 g/dL), thrombocytopenia (Platelets: 57.000), low white blood cell count (WBC: 2800), acute kidney failure (serum creatinine level was 506 µmol/L, uremia: 43 mmol/L), low prothrombin level (25%), elevated lactate dehydrogenase (696 UI/L), high lactatemia (4.6 mmol/L), elevated pro BNP (5800 ng/L), elevated troponin (162 µg/L), mild hepatic cytolysis (SGOT:149 UI/L, SGPT: 49 UI/L), high ferritinemia level:1983 ng/mL, low serum albumin: 25g/L, high triglyceride level 2,53 g/L. Blood smear showed Schistocytes, with normal Haptoglobin: 4g/L. Thoracoabdominopelvic CT scan showed: micronodules of the lungs and a splenic infarction. Electrocardiogram displayed tachycardia with episodes of atrial fibrillation. Heart ultrasound showed no signs of endocarditis. Left ventricular ejection fraction was normal. After fluid resuscitation, high doses of noradrenalin were needed in order to restore conve-

nient blood pressure (9 mg per hour of noradrenaline). The patient was prescribed broad-spectrum antibiotics, antiviral drugs. Intermittent kidney replacement was also needed. On third day of symptoms, fever kept tray (39°-40°C), blood pressure was low with high doses of noradrenalin along with kidney failure. Blood, sputum and urine cultures were all negative for bacteria. Viral infection was ruled out via viral panel. SARS-CoV-2 PCR of nasopharyngeal swab was negative. Blood screening for CMV, aspergillosis, pneumocystosis, hepatitis and HIV were all clear. Invasive Candidiasis was also ruled out.

Further interrogation revealed a history of uncomplicated Covid 19 infection detected with PCR of nasopharyngeal swab a month ago. Hence, multisystem inflammatory syndrome in adults was suspected. Accordingly, the patient was prescribed intravenous immunoglobulin during three days (0.4g/kg/day) as well as high doses of Methylprednisolone (1 g/day) for three days followed by tapering. Two days after the initiation of treatment, fever dropped down, hemodynamics improved allowing noradrenaline with drawal, urine output increased, and blood creatinine decreased (serum creatinine level: 116 µmol/L, uremia: 18 mmol/L, inflammatory markers decreased (CRP: 20 mg/L), but low blood count persisted (Hb:7.1 g/dL, WBC: 500, neutrophil count: 400, Platelets: 26.000). Bone marrow puncture revealed an hemophagocytosis, the patient was prescribed Folate and Vitamin B 12 in adjunction with corticosteroids. After 11 days of stabilization, the patient developed a septic shock due to low neutrophil count, and died shortly after.

Discussion

SARS-CoV-2 can trigger a range of inflammatory syndromes. This serious hyper inflammatory condition presents approximately 4 weeks after onset of acute Covid-19 with extrapulmonary multi organ dysfunction [5]. In our case, the onset of the symptoms is exactly one month after confirmed Covid 19 infection.

The virulent strain of SARS-CoV-2 appears to cause a post-infectious inflammatory syndrome similar to Kawasaki disease in pediatric population [6]. After SARS-CoV-2 infection, Kawasaki-like multisystem inflammatory syndrome can also occur among adults (MIS-A) [7]. Typically, these patients had high-grade fever, severe asthenia, abdominal pain and diarrhea, hypotension related to cap-

illary leak syndrome and vasoplegia, and pronounced biologic inflammatory syndrome [4].

In our case, after ruling out possible infection, given the patient's constellation of signs (fever for more than 5 days, erythema multiforme-like rash, bilateral non-exudative conjunctivitis, erythema or cracking of the lips), he matches the American Heart Association (AHA) criteria for Kawasaki disease [8]. Subsequently, he was diagnosed with Kawasaki-like multisystem inflammatory syndrome associated with COVID-19. Given low systolic blood pressure requiring vasoactive drugs, the patient presented with Kawasaki Disease Shock Syndrome [9]. It's important to highlight that the patient had no active Covid 19 infection at the time of diagnosis.

In some cases, patients diagnosed with after Covid MIS-A show prominent cardiac dysfunction [10], some of them show signs of severe myocarditis [4]. In our case, the patient had rapid atrial fibrillation, with elevated pro BNP (5800 ng/L), high troponin level (162 µg/L). But transthoracic heart ultrasound displayed normal left ventricular function.

In our case, splenic infarction found on CT scan can be correlated to Kawasaki disease's angiitis [11]. As for acute kidney failure in MIS-A, it is thought to be mainly related to cytokine-mediated hypotension and cardiac dysfunction, leading to renal hypoperfusion [12]. However, kidney thrombotic microangiopathy have been reported in some cases [7]. In our case, in addition to high uremia and high serum creatinine level, Schistocytes were found on blood smear with no proteinuria.

Although the cause of Kawasaki disease remains unknown, the most widely accepted theory is an aberrant immune response to an infectious trigger [10]. Emerging reports depict the phenotype of MIS-C as a combination of Kawasaki disease, toxic shock syndrome, and macrophage activation syndrome [10]. In our case, macrophage activation syndrome was confirmed via bone marrow puncture. The resolution of symptoms and normalization of vital signs within two days after treatment with corticosteroids and intravenous immunoglobulin supports the diagnosis. Aspirin was not prescribed given the patient had severe thrombocytopenia.

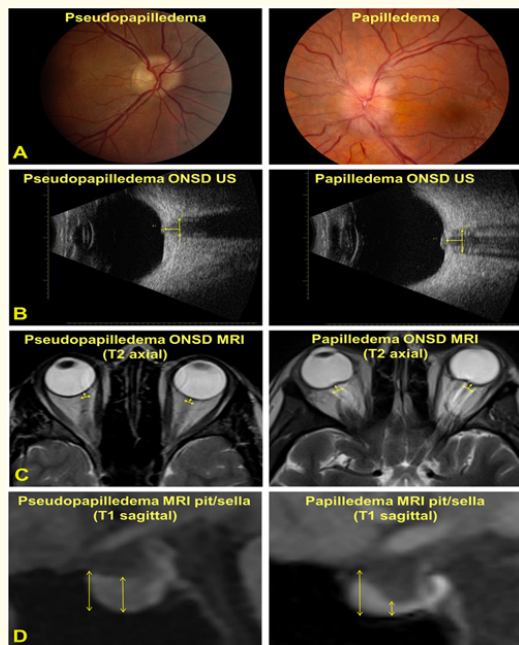


Figure 1: Optic nerve papilledema and drusen on fundus oculi (A), ultrasound (B) and MRI (C-D).

Conclusion

Multisystem inflammatory syndrome in adults (MIS-A) is a delayed complication due to Covid 19 infection. In fact, distinguishing MIS-A from other severe infectious or inflammatory disease is challenging. We report a rare case of multisystem inflammatory syndrome with Kawasaki like Disease associated with Kawasaki Shock Syndrome and macrophage activation syndrome. Despite being rare, this clinical presentation requires urgent recognition, hemodynamic support, and immediate management.

Bibliography

1. Hamann S., et al. "Optic disc drusen: understanding an old problem from a new perspective". *Acta Ophthalmologica* 96.7 (2018): 673-684.
2. Malmqvist L., et al. "Progression Over 5 Years of Prelaminar Hyperreflective Lines to Optic Disc Drusen in the Copenhagen Child Cohort 2000 Eye Study". *Journal of Neuroophthalmology* 40.3 (2020): 315-321.
3. Chang MY and Pineles SL. "Optic disc drusen in children". *Survey of Ophthalmology* 61.6 (2016): 745-758.
4. Khonsari RH., et al. "Drusen de la tête du nerf optique ou œdème papillaire? [Optic disc drusen or true papilledema?]". *Revue Neurologique (Paris)* 166.1 (2010): 32-38.

5. Brémond-Gignac D and Milazzo S. "Drusen du nerf optique de l'enfant et complications hémorragiques [Drusen of the optic nerve in children and bleeding complications]". *Journal Français d'Ophtalmologie* 34.9 (2011): 679-82.
6. Chang MY and Pineles SL. "Optic disk drusen in children". *Survey of Ophthalmology* 61.6 (2016): 745-758.
7. Noval S., et al. "Visual field defects due to optic disk drusen in children". *Graefe's Archive for Clinical and Experimental Ophthalmology* 251.10 (2013): 2445-2450.
8. Yan Y and Liao YJ. "Updates on ophthalmic imaging features of optic disc drusen, papilledema, and optic disc edema". *Current Opinion in Neurology* 34.1 (2021): 108-115.
9. Harraka P., et al. "Increased retinal drusen in IgA glomerulonephritis are further evidence for complement activation in disease pathogenesis". *Scientific Report* 12.1 (2022): 18301.
10. Sibony PA., et al. "Optical Coherence Tomography Neuro-Toolbox for the Diagnosis and Management of Papilledema, Optic Disc Edema, and Pseudopapilledema". *Journal of Neuroophthalmology* 41.1 (2021): 77-92.
11. Schwartz R., et al. "A Deep Learning Framework for the Detection and Quantification of Reticular Pseudodrusen and Drusen on Optical Coherence Tomography". *Translational Vision Science and Technology* 11.12 (2022): 3.
12. Gise R., et al. "Diagnosis and Imaging of Optic Nerve Head Drusen". *Seminar on Ophthalmology* 34.4 (2019): 256-263.
13. Floyd MS., et al. "Measurement of the scleral canal using optical coherence tomography in patients with optic nerve drusen". *American Journal of Ophthalmology* 139.4 (2005): 664-669.
14. Thompson AC., et al. "Bruch's membrane opening on optical coherence tomography in pediatric papilledema and pseudopapilledema". *Journal of AAPOS* 22.1 (2018): 38-43.e3.
15. Malmqvist L., et al. "Long-term evolution of superficial optic disc drusen". *Acta Ophthalmologica* 95.4 (2017): 352-356.
16. Yi K., et al. "Imaging of optic nerve head drusen: improvements with spectral domain optical coherence tomography". *Journal of Glaucoma* 18.5 (2009): 373-378.
17. Murthy RK., et al. "In-vivo high resolution imaging of optic nerve head drusen using spectral-domain Optical Coherence Tomography". *BMC Medical Imaging* 10 (2010): 11.
18. Pineles SL and Arnold AC. "Fluorescein angiographic identification of optic disc drusen with and without optic disc edema". *Journal of Neuroophthalmology* 32.1 (2012): 17-22.