

Case Report

Sarcoidosis in a Toddler: A Rare Presentation

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Abstract:

Sarcoidosis is rare multi-system granulomatous inflammatory disorder. It is reported to present by two distinct presentations in childhood. Sarcoidosis presents in the second decade of life by pulmonary infiltration, lymphadenopathy and hepatosplenomegaly, and rarely before the age of 4 years by typical triad of uveitis, rash and arthritis. We report a child 1 year and 9 months with sarcoidosis who presented by one month fever, hepatosplenomegaly, uveitis, pulmonary infiltration and elevated levels of angiotensin-converting enzyme. Chest computerized tomography revealed multiple enlarged mediastinal and multiple hilar lymphadenopathy. A right inguinal lymph node biopsy showed non-caseating granuloma. Other etiologies were investigated for and ruled out. Hence, sarcoidosis can present in children before 4 years with uveitis, hepatosplenomegaly, pulmonary infiltrates, mediastinal and hilar lymphadenopathy, in the absence of rash and arthritis. Sarcoidosis in children is rare and clinical spectrum can vary considerably. Diagnosis of sarcoidosis should be suspected in any child with uveitis.

Level of Evidence of Study: IV (1).

Keywords: sarcoidosis; uveitis; rash; pulmonary infiltration; hepatosplenomegaly; infantile sarcoidosis.

Abbreviations: ACE: angiotensin converting enzyme; CT: computerized tomography.

Introduction

Sarcoidosis is a multi-system inflammatory non-caseating and non-necrotizing granulomatous disease that is rare in children. In adults it presents acutely by fever, cough, dyspnea, anterior uveitis, or as an insidious onset disease with cough and hilar lymphadenopathy (2). It is a diagnosis of exclusion. Sarcoidosis is very rare in children. It is reported to have two distinct clinical presentations in children. The presentation before the age of 4 years is the rarer, and presents by a triad of uveitis, arthritis and rash. It is named congenital sarcoidosis; Blau syndrome (3). The presence of epithelioid cells and multinucleated giant cells on a skin, synovial or conjunctival biopsy is diagnostic (4). During the second decade, children present by the typical adult presentation of pulmonary infiltrates, hilar and mediastinal lymphadenopathy, and hepatosplenomegaly.

The purpose of this work was to report the adulthood phenotype presentation of sarcoidosis disease in a toddler.

Case Presentation

A 1 year and 9 months old boy presented by history of fever for one month. There was no history of oral ulcers, rash, arthritis, gastrointestinal tract symptoms, renal symptoms or other system affection. There was no family history of similar condition in the family. By examination the child weight and height percentiles were within the 50th percentile for his age. Abdominal examination revealed hepatosplenomegaly and lymphadenopathy. He had bilateral granulomatous panuveitis, with keratic precipitates, posterior synechia and complicated cataract, and by ocular ultrasound; there was evidence of vitritis with left starting atrophica.

(Figures 1-3). Non contrast computerized tomography (CT) chest revealed: few enlarged mediastinal lymph nodes about 1.3 cm in cross sectional dimensions while post contrast CT chest revealed: multiple enlarged mediastinal lymph nodes and multiple enlarged hilar lymph nodes and right para sternal lymph node.

His post contrast CT abdomen revealed: enlarged size of liver and spleen and multiple enlarged abdominal (para aortic, aorto-caval, mesenteric, celiac and portocaval) lymph nodes. Tuberculin test was negative despite being vaccinated early in life. Biopsy from right inguinal lymph node showed: non –caseating granulomatous lymphadenitis. Lab testing confirmed high level of angiotensin converting enzyme (ACE enzyme). His serum calcium level was within normal. Antinuclear antibody and rheumatoid factor were negative. Echocardiography detected no abnormality. Bone marrow aspirate and bone marrow biopsy demonstrated no abnormality. Investigations for underlying immunodeficiency were negative. He received pulsed steroids 30mg/kg /day for 3 days followed by oral steroids that controlled the fever and uveitis. Left eye vision was compromised. Methotrexate was added 2 weeks later. Right eye developed uveitis, that responded to periocular corticosteroid injections.

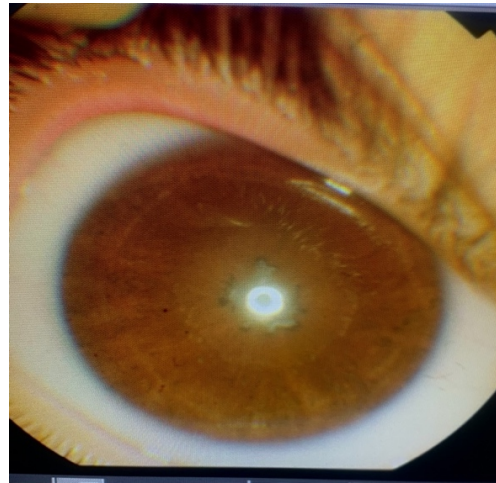


Figure 1: Anterior segment photo of the right eye- showing a washed-out iris pattern, posterior synechia (secclusio pupillae) and complicated cataract due to chronic uveitis.

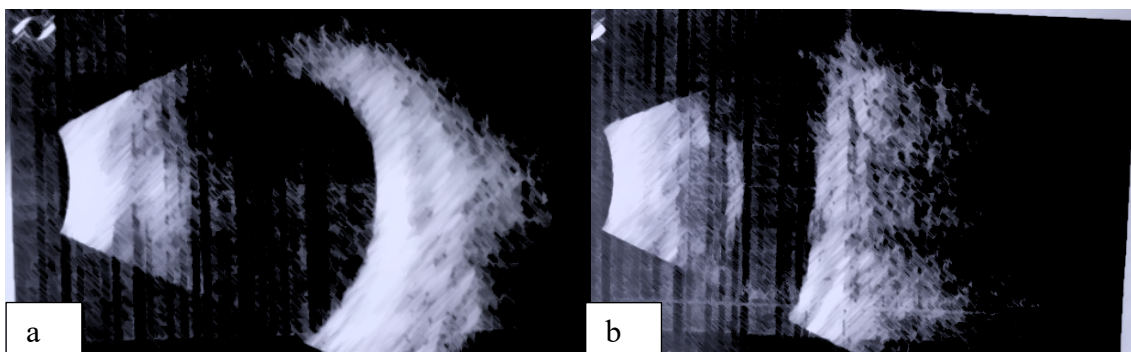


Figure 2: Ocular ultrasound of right (a) and left eye (b).

Right ultrasound shows a normal contour with vitreous floaters (vitritis). Left eye also shows evidence of vitritis with a flattened contour with and an elevated optic nerve head due to hypotony.

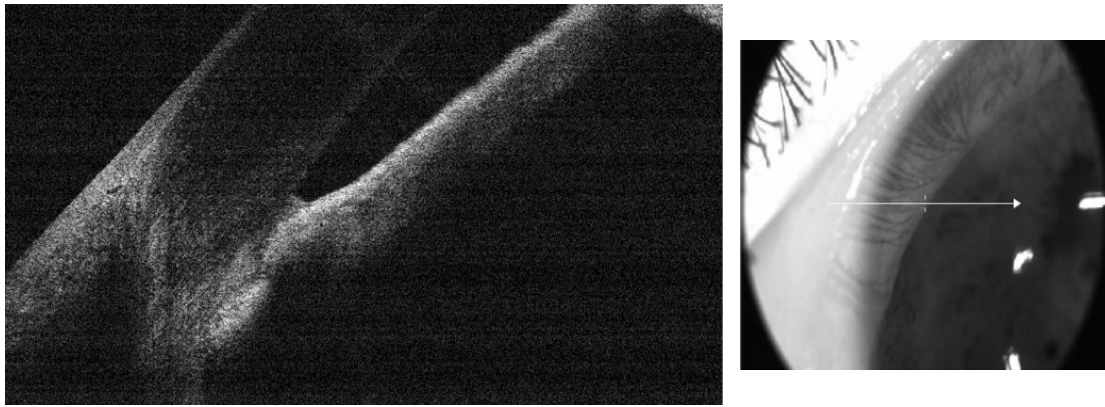


Figure 3. Anterior segment optical coherence tomography of the left eye showing starting peripheral anterior synechia.

Discussion

Sarcoidosis is a very rare disease, that has different clinical picture and organ involvement according to age at presentation. The child we describe proves that sarcoidosis in early childhood is not limited to the triad of Blau syndrome; namely: uveitis, arthritis and rash, and the clinical spectrum of sarcoidosis might be variable (6). There are a few studies that aim to describe age or severity related presentations. We report a different presentation; a young toddler presented with fever, hepatosplenomegaly, generalized lymphadenopathy, and uveitis that proved to be sarcoidosis. At the time of diagnosis there was no rash, arthritis, hypercalcemia, renal involvement or neuro-sarcoidosis. Despite lack of cough and other symptoms suggestive of lung disease, chest CT revealed pulmonary infiltrates and hilar lymphadenopathy. The overlap of clinical presentation affirms that sarcoidosis in early childhood should be suspected in any child with uveitis. Angiotensin-converting enzyme (ACE) can be significantly elevated and useful as a marker of disease activity, but the test is not specific for sarcoidosis (5). It is important to search for pulmonary involvement in any child suspected to have sarcoidosis even in absence of symptoms.

Untreated uveitis in sarcoidosis results in permanent visual affection. Uveitis in children might be associated with juvenile idiopathic arthritis (7), lymphoma (8), enthesitis-related arthritis, Behçet syndrome, tubulointerstitial nephritis, infections (7), immunodeficiency (9) and drug induced (10). Detailed history, clinical examination and relevant investigations is necessary to reach a diagnosis (11). Granulomas in pediatric age are rare, but might be caused by sarcoidosis, granulomatous angiitis, Crohn's disease, tuberculosis and chronic granulomatous disease of childhood (12) among other rare causes (11). Management is directed at the cause, and despite the diversity of investigations that are necessary to reach a diagnosis, the uveitis is an urgent pressing factor that warrants expediting the investigations. Sarcoidosis is a diagnosis of exclusion. Lymph node biopsy remains the mainstay of diagnosis. Uveitis and granulomas in toddlers and childhood are rare but might the presenting sign of very serious disease and untimely intervention might be complicated by loss of vision.

Conclusion

Sarcoidosis should be ruled out in any child with uveitis even in the absence of rash and arthritis. Sarcoidosis is a diagnosis of exclusion, hence, all other causes should be ruled out prior to initiation of treatment. Sarcoidosis clinical presentations are variable in childhood.

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CONFLICT OF INTEREST

The authors declare no conflict of interest. Authors declare veracity of information.

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