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Research Article

An Analysis of Early Onset Sarcoidosis and Pulmonary Findings at Presentation

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Abstract

Early onset sarcoidosis (EOS) is usually defined as an early presentation of the disease which is devoid of pulmonary features. Children are diagnosed on or before 5 years of age, and may present with some or all of a triad of classic features: arthritis, rash and uveitis. The purpose of this study was to analyze the literature in order to identify cases of EOS with pulmonary involvement, during the early course of the disease.

Keywords: *Early Onset Sarcoidosis; Pulmonary; Arthritis*

Introduction

Early onset sarcoidosis (EOS) is an early presentation of the disease with lifelong clinical progression. The features of the disease include a classic triad of clinical disease i) arthritis, ii) dermatitis, iii) uveitis. The differential diagnosis of the disease includes systemic onset JRA. The rash of EOS is distinct from JRA and typically is described as papular and/or scaling. EOS patients may have synovial thickening and effusions of the synovium without limitation of movement, but some children do have destructive arthritis. EOS is not currently defined to involve the pulmonary system [1-4].

The purpose of this study was to analyze the literature to identify cases of EOS with pulmonary involvement, to test the hypothesis that a form of more aggressive EOS may exist which does involve the respiratory tract.

Methods

Literature Search

The PubMed databases were searched using the MeSH terms 'early onset sarcoidosis' and 'lung' OR 'pulmonary' to identify literature which may describe EOS cases.

Identification of Cases

Once the citations were identified, articles were included if they described EOS cases or patients.

The EOS cases were included for further analysis if:
i) Cases of EOS defined as diagnosis at 5 years of age or younger were identified in the article or references of the article, ii) Cases were included regardless of clinical description of pulmonary findings. The exclusion criteria were: i) older age at presentation, ii) non-sarcoidosis diagnosis, ii) non-English literature.

Further case analysis of respiratory clinical, imaging and diagnostic features among these EOS patients was performed to describe the presence of early pulmonary findings. Pulmonary findings were defined as any symptom, sign or testing consistent with respiratory disease present with EOS at or before the age of five years.

The findings included for analysis included: i) age at diagnosis, ii) clinical presentation, iii) pulmonary clinical disease, iv) imaging, v) biopsy consistent with sarcoidosis, vi) pulmonary function testing, vii) mutational analysis as available.

Results

There were a total of 16 articles, meeting the inclusion and exclusion criteria identified in the literature search [1-16]. The total number of EOS cases was 106, and among these 20 (18.8%) had pulmonary findings at or earlier than five years of age. The early pulmonary findings included a number of lines of supporting evidence. The articles identified patients with biopsies consistent with sarcoidosis, but not all of the studies analyzed this information. The most common presentation of EOS was a rash e.g. papular rash at the time of diagnosis.

The radiographic studies for the cohort included plain chest radiographs and HRCT of the thorax. Among the findings were alveolar and interstitial consolidation with and without hilar adenopathy, pleural disease, pneumonia and one patient with a right hydrothorax.

The clinical presentations of respiratory disease included the following symptoms and signs: hypoxia, tachypnea, lower respiratory tract infections and hydrothorax.

The pathologic diagnosis did not require lung biopsy in all except one case.

Discussion

The purpose of this study was to identify whether there are cases in the literature of EOS and pulmonary disease. Classically, EOS has been recognized as a presentation of childhood sarcoidosis which does not involve the respiratory system. Patients typically have one or more feature of a classic triad of skin, ocular and rheumatologic findings. This study identified a sub-group of EOS patients with pulmonary disease. This may represent a more aggressive form of Sarcoidosis in which the progression is accelerated. EOS patients therefore should be assessed for the presence of pulmonary disease, and an imaging study may be useful in this regard.

There was one case of an 11-month-old patient with sarcoid like biopsy confirmed pulmonary lesions, which was proposed as a case of infant sarcoidosis. The child did have HRCT findings of both alveolar and interstitial consolidation following a Parainfluenza III positive nasal swab and respiratory distress. Some of the radiographic findings may be attributed to infection, but the biopsy performed was determined to be consistent with sarcoid like granulomatous disease. This case raises the possibility that a lower respiratory tract infection may lead to sarcoid like granulomatous disease and serve as a triggering event for the pathologic findings [9].

Other diagnostic testing to consider in the evaluation of these patients would include early skin biopsy to establish the diagnosis and infant pulmonary function testing to detect early lung function deficits. Based on a database review of findings among Childhood Sarcoidosis patients, imaging showed bilateral hilar adenopathy in 40% of the patients [7]. In another study, HRCT was positive in 95% of children with sarcoidosis. Pulmonary function testing among these patients were variably abnormal and often showed normal DLCO measurements among younger patients [1].

Other considerations in the analysis of EOS patients is the possible presence of NOD2 mutations. The differential includes Blau Syndrome, which shares a common presentation with EOS. Blau syndrome consists of early onset granulomatosis, arthritis, uveitis and rash. The disease unlike EOS is inherited in an autosomal dominant manner. Kanazawa., *et al*. performed an analysis of ten EOS patients to identify CARD15 mutations which have also been identified among Blau syndrome families. Among the 10 EOS patients studied, 9 had heterozygous CARD15 missense mutations [8].

Childhood sarcoidosis is considered distinct from EOS since it most commonly affects older children and adults. The common presenting features include hilar adenopathy, pulmonary infiltrates, ocular and skin disease.

Given the findings of this study, clinical management of young children with EOS should include performance of a thoracic HRCT scan to determine whether there is respiratory tract involvement.

Based on this analysis, a novel presentation of EOS includes pulmonary disease is reported for the first time in an analysis of the literature, and this should be further investigated in future studies of pathogenesis of the disease.

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