



POSTER PRESENTATION

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Clinical manifestations of four patients diagnosed with early-onset sarcoidosis or sarcoid-like syndrome

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Introduction

Sarcoidosis is a rare multisystemic granulomatous disease. Pulmonary involvement is common in adults, but any organ can be affected.

Objectives

To describe the main features of 4 pediatric patients diagnosed with early-onset sarcoidosis (EOS) or sarcoid-like syndrome.

Methods

Medical charts were reviewed.

Results

Four patients were enrolled, 2 female (cases 1 and 2) and 2 male (cases 3 and 4). Their main characteristics are shown in table 1. The age at disease onset was 1.5, 10, 0.6 and 11 years respectively. The time until diagnosis ranged from 4 days to 8 years. The two cases with EOS (1 and 3) had been previously diagnosed as Juvenile Idiopathic Arthritis. Both started at a short age with the classical triad and carried a heterozygous gain-of-function *NOD2* mutation. Patient 2 seemed to be a late-onset sarcoidosis but persistent hypogammaglobulinemia and poor antibody production suggested a CVID, despite she did not suffer

Table 1 Main features of the four enrolled children

| | Clinical data | Analytical data | ACE | Non-caseating granulomata | Diagnosis |
|---|---------------------------------------------------------------------------|-------------------------------------------------------------------------------------|------|---------------------------|-------------------------------|
| 1 | Arthritis, rash, uveitis | Heterozygous p.R334Q Mutation at <i>NOD2</i> gene | 27.5 | Skin | EOS |
| 2 | Splenomegaly, lymphadenopathy, interstitial/nodular lung disease | Lymphopenia Thrombocytopenia Hypogammaglobulinemia Poor antibody responses | 92 | Lung | CVID Sarcoid like syndrome |
| 3 | Arthritis, rash, uveitis | Heterozygous p.C495Y Mutation at <i>NOD2</i> gene | 19 | Skin Synovial Liver | Blau's syndrome |
| 4 | Right facial paralysis, parotid enlargement, uveitis, high fever | | 74.8 | | Heerfordt's syndrome |

ACE = angiotensin converting enzyme serum level (normal 10-50 IU/l)

CVID = common variable immunodeficiency

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from recurrent infections. Other granulomatous lung diseases were dismissed in this case. Finally, diagnosis in patient 4 was made according to his clinical manifestations and the slight increase in ACE level.

Conclusion

Diagnosis of sarcoidosis in pediatric patients is often delayed because the disease is not suspected. Pulmonary involvement occurs less frequently in pediatric than in adults patients. This condition requires multidisciplinary management.

Disclosure of interest

None declared.

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