CASE REPORT

Distal Arthrogryposis Type Six and Systemic Lupus Erythematosus, in a Girl: First Pediatric Case

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Abstract

Arthrogryposis type six is one of the distal arthrogryposes. It associates arthrogryposis of the hands and sensorineural deafness. Male-to-male transmission was observed. No gene has been identified to date. We report a unique case of a girl presenting with congenital arthrogryposis-like hand anomaly, sensorineural deafness, and acute onset of systemic lupus erythematosus (SLE).

Introduction

Arthrogryposis-like hand and deafness syndrome or distal arthrogryposis type 6 (DA6) is characterized by an arthrogryposis-like hand anomaly and a sensorineural deafness. It has been described in one family. Transmission occurs between male individuals. We describe a case of DA6 in a six-year-old girl who also had an acute onset of systemic lupus erythematosus (SLE). This case is –to our knowledge- the first to be described to date.

Case Report

The proband, product of a first-degree consanguineous marriage, a girl, with no family history of deformities, hearing loss, or auto-immune disease, who was born with arthrogryposis of the two hands, and had congenital deafness. Audiometric tests revealed a sensorineural deafness; after which, a cochlear implant was placed at two-year-old. As for her hand contractures, no treatment was applied because of family refusal. Fortunately, the child could cope with daily living needs. The girl developed a mild mental retardation on follow up. At the age of six-years, she had inflammatory polyarthralgia of big joins, treated symptomatically with anti-inflammatory medication with no improvement. Three months later, an acute onset of a cutaneous rash alarmed the parents. At examination, she had on top of bilateral hands arthrogryposis, a generalized discoid rash sparing the palms, and the soles of the feet. She had no other deformities. The parents reported photosensitivity and excessive hair loss. Markers of inflammation were very high (ESR = 76 mm the 1st hour). We found leucopenia (WBC = 3400/mm³), normochromic normocytic anemia (HB = 8 g/dl, MCV = 85 fl, MCHC = 23 pg) with normal reticulocytes (RET = 104000/mm³), and no thrombocytopenia on blood count (was there thrombocytopenia?).

Cutaneous biopsy was performed, and histology confirmed the diagnosis of acute cutaneous lupus erythematosus. Screening for a cardiac, pulmonary, nephrological, neurological and ocular involvement was negative. Immunological testing was positive to anti-nuclear antibodies, anti-ds DNA antibodies. Meanwhile, complement, rheumatoid factor, anti-Sm antibodies, anti-Rnp antibodies were negative. Screening for antiphospholipid and Gougerot-Sjögren syndromes was negative. We started oral steroids (prednisone) at 15 mg/day and Hydroxychloroquine at 6 mg/kg/day, as well as a pediatric sunscreen. After two weeks, there was no more arthralgia and total remission of cutaneous symptoms. Steroid tapering was started three months later. After twelve months, the child was still asymptomatic;
Conclusions

Acute SLE in the course of DA6 could be easily mistaken as Rhupus, especially if it’s a girl. Congenital onset of the hands’ deformities, as well as the discovery of a hearing loss can rectify the diagnosis. Unavailable local facilities did not allow further investigations of this unique case.

Contribution

HN: Writing the paper, RQ, AB, IA: Final approval.

Conflicts of Interest

All authors declare that they have no conflict of interest.

References