



Farber Disease - A Case Report

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Abstract

Farber Disease is a rare autosomal recessive disorder that results from the deficiency of the lysosomal enzyme acid ceramidase leading to accumulation of ceramide in various tissues especially joints. Symptoms may begin in the 1st year of life with painful joint swelling and nodule formation, which is sometimes diagnosed as polyarticular JIA. We report a case of 3month old male baby presented to us with painful swelling of multiple joints and contractures since birth. For rarity of the case and to create awareness among the physician regarding this JIA mimic condition.

Keywords: Farber disease, Ceramidase, joint swelling, JIA.

Introduction

FD is an inherited autosomal recessive lysosomal storage disorder due to deficiency of lysosomal acid ceramidase which causes accumulation of ceramide within different tissues leading to abnormalities in joints, larynx, liver, CNS and other organs.¹ It is named after Sydney Farber in 1957. Also known as Fibrocystic dysmucopolysaccharidosis / disseminated lipogranulomatosis.² It usually starts as early as 1st week of life with painful joint swelling and subcutaneous nodule formation, joint deformity and hoarseness of voice with breathing difficulty. In some patients it may present with hepatosplenomegaly, moderate to severe CNS, heart and lung involvement.³ Depending on the severity and clinical manifestations seven phenotypes have been identified.⁴ Most children with FD die by 2 years usually from the lung disease. Diagnosis is mainly confirmed by measuring the acid ceramidase level or

histopathological finding in subcutaneous nodule biopsy. These finding includes presence of granulomatous inflammation along with foam cells and fibrovascular stroma.

In typical case of FD the triad of Subcutaneous nodules joint and laryngeal involvement is sufficient to diagnosed the case. Hematopoietic stem cell transplantation [HSCT] can be used to treat the patients without CNS involvement.⁵

Case Detail

A 3month old 1st order male baby born out of 3rd degree consanguineous marriage presented to us with painful swelling of multiple joints & deformity since birth with history of repeated hospitalization for ARI.

Examination revealed normal anthropometry with mild motor delay. His vitals parameters were within normal limits. Musculoskeletal system revealed severe pain, multiple joints involvement mainly interphalangeal with restricted movement

and contracture. Thickened skin and subcutaneous tissues with hyperpigmented nodular lesions over knuckles and ankles were seen. On systemic examination hepatosplenomegaly was there, other system were normal.

Investigations revealed Hb 12gm/dl and TLC 29000/cmm with neutrophilic leucocytosis. TPC was normal. Blood biochemistry showed normal

RFT, LFT and Electrolytes. X-ray of wrists, elbow showed evidence of osteopenia, swelling and deformity. 2D ECHO was normal. Ceramidase level estimation and genetics study could not be done. On the basis of presentation and clinical finding provisional diagnosis was made.



Fig-1 X-ray wrist (L) showing contracture and deformity



Fig-2 Swelling of wrist and ankle joint with hyperpigmented nodular lesion.

Discussion

FD is very rare, about 80 cases have been reported. The clinical presentation of Farber disease consists of joint pain and swelling with subcutaneous nodules in the vicinity of joint or over the pressure point. These affected joints are very painful and lead to progressive joint stiffness, restriction of movements by contractures and finally to deformity.

Hepatosplenomegaly occurs in 25% of the cases. Depending on severity and manifestation seven different phenotypes have been identified. Our patient is mostly fitting to type 1 classical variety without CNS involvement. In a typical case triad of joint involvement, subcutaneous and laryngeal nodule is enough to make clinical diagnosis.

Conclusion

The diagnosis of this rare disorder should be suspected in patients who have painful joints and nodule formation presenting at early part of infancy which closely mimic polyarticular JIA and hand foot syndrome of SCD.

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