CASE REPORT

Disseminated Lipogranulomatosis

RK MONDAL, M NANDI, S DATTA AND M HIRA*

*From the Department of Pediatric Medicine and *Department of Orthopedics, IPGMER and SSKM Hospital, Kolkata, India.*

Corrspondence to:
Dr Madhumita Nandi, 6/6, Naren Sarkar Road, Barisha, Kolkata, West Bengal 700 008, India. *E-mail: madhumitabanik@rediffmail.com*
Manuscript received: February 11, 2008; Initial review completed: March 7, 2008; Revision accepted: March 24, 2008.

Farber disease or disseminated lipogranulomatosis is a rare inherited disorder of lipid metabolism resulting from a defect in ceramide degradation. Because of the feature of nodular swellings around various joints, this may sometimes be confused with juvenile idiopathic arthritis. We report a 4-year-old boy with Farber disease who presented with nodular swellings around the joint, angle of the mouth and conjunctiva, and was subsequently diagnosed to be a case of Farber Disease.

Keywords: *Farber disease, Lipogranulomatosis.*

Farber disease is a rare inherited autosomal recessive disorder of lipid metabolism in childhood population due to a genetically determined defect in ceramide degradation.

The main importance of reporting this rare disease is that it may be confused sometimes with polyarticular juvenile idiopathic arthritis(JIA).

CASE REPORT

A 4-year old boy was referred to us as a case of polyarticular JIA with joint pain, deformity and nodular swelling around multiple joints, and nodules at the angle of the mouth and conjunctiva since childhood (***Fig.* 1,2**). The child also had difficulty in walking. He had also developed hoarseness of voice for last two years. There was no history of skin rash, convulsion, bleeding diathesis etc. His developmental milestones were within normal limits. His two other siblings were normal.

Examination revealed weight of 8.5 kg (expected 16 kg), height 80 cm (expected 100 cm) and head circumference 46.5 cm (expected 48 cm). His vital parameters and general physical examination were within normal limits. Musculoskeletal system revealed multiple soft to hard nodular irregular tender swelling around multiple joints with deformity and limitation of movement. Direct laryngoscopy revealed multiple laryngeal granulomas in both epiglottic and supraglottic area. These were the cause of hoarseness of voice.

Investigations revealed hemoglobin of 11.5g/dL and TLC 7000/cmm (P72, L22 and E4). Platelet count was normal. Blood biochemistry showed albumin 2.8g/dL, alkaline phosphatase 260 IU/L and SGPT 40 IU/L. His blood glucose, blood pH, renal function and electrolytes were within normal limit. Routine urine examination, Mantoux test, and chest X-ray did not reveal any abnormality. *X*–ray of shoulder joint revealed calcification of axillary lymphnode on left side (***Fig.* 3**). *X*–ray of wrists, knee and ankle showed generalized osteopenia with juxtra-articular osteopenia. Rheumatoid factor and anti-nuclear factor assay were normal. Biopsy of the nodular swelling revealed highly cellular tissue with scattered large foamy cells having round to oval nuclei and granular eosinophilic cytoplasm suggestive of storage cells. Ceramidase level estimation and genetic studies could not be done. On the basis of clinical presentation and investigation findings, a diagnosis of disseminated lipogranulomatosis or Farber disease was made.

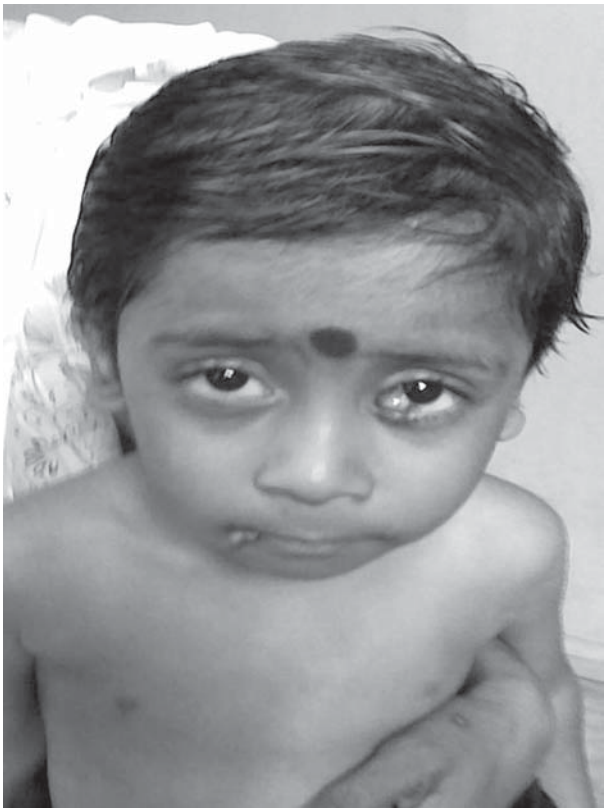

FIG. 1 *Clinical photograph of the patient of Farber's disease showing nodular swelling at the angle of the mouth and in left conjunctiva.*

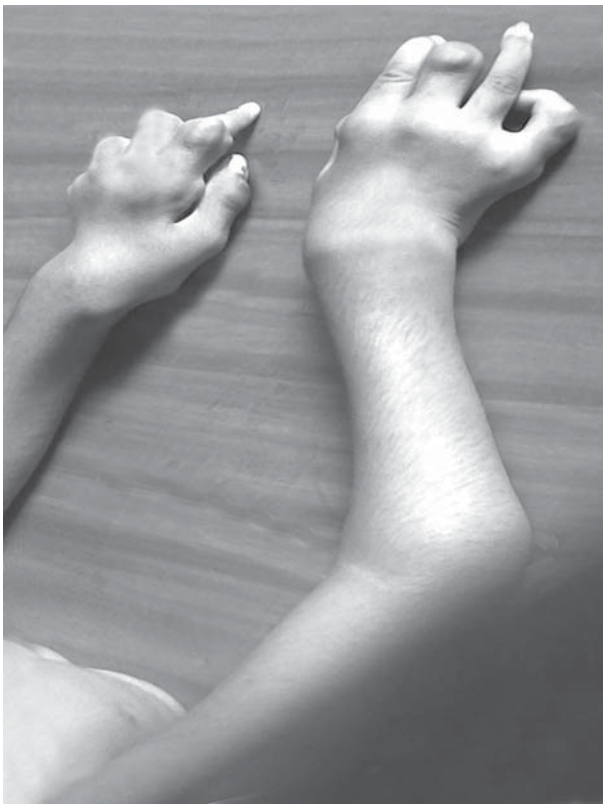

FIG. 2 *Clinical photograph showing deformity and nodular swelling around the wrists and elbow joints.*

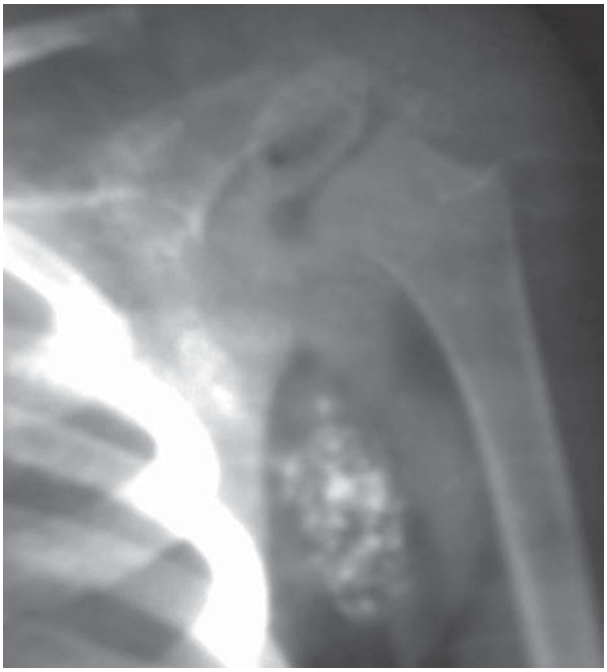

FIG. 3 *X-ray Shoulder joint showing lymph node calcification in the axilla.*

DISCUSSION

Features of Farber disease include hoarseness of voice, joint swelling, sub-cutaneous nodules etc. Lipid analysis and pathological investigation confirm the diagnosis by showing the presence of ceramidase deficiency characteristic for Farber disease(1). This disease is usually lethal within first two decades of life(2). Familial lipogranulomatosis (Farber disease) has been rarely reported in world literature(3-5) and probably only two such cases were reported from India before(6,7).

Patients with Farber disease are sometimes misdiagnosed as cases of juvenile idiopathic arthritis (JIA), as happened in the index case. Very early onset, extensive nodule formation around joints, nodular deposits of lipogranuloma in other sites like conjunctiva, and hoarseness of voice due to laryngeal granuloma *etc* help to differentiate it from JIA. Prenatal diagnosis can be done by detection of ceramidase level in chorionic villus sampling(8).

Contributors: RKM, MN, SD and MH were involved in diagnosis and management of the case. MN drafted the paper and searched the literature. RKM, SD and MH contributed to paper writing. All authors approved the final manuscript.

Funding: None.

Competing interests: None stated.

REFERENCES

1. Ozaki H, Mizutani M, Hayashi H, Oka E, Ohtahara S, Kimoto H, *et al*. Farber disease (disseminated lipogranulomatosis): the first case reported in Japan. Acta Med Okayama 1978; 32: 69-79.
2. el Sharkawy L, Abdallah H, Marzouk S. Farber disease: a cause of hoarseness of the voice in children. Rev Laryngol Otol Rhinol 2000;121: 261-265.
3. Tanaka T, Takahashi K, Hakozaki H, Kimoto H, Suzuki Y. Farber disease (disseminated lipogranulomatosis)-a pathological, histochemical and ultrastructural study. Acta Pathol Jpn 1979; 29 : 135-155.
4. Zetterstrom R. Disseminated lipogranulomatosis (Farber disease). Acta Paediatr 1958; 47: 501-510.
5. Sugita M, Dulaney JT, Moser HW. Ceramidase deficiency in Farber disease (lipogranulomatosis). Science 1972; 178: 1100-1102.
6. Singhi PD, Padmini P, Mantri SB. Farber disease. Indian Pediatr 1991; 28: 1338-1341.
7. Chandwani R, Kuwar AS. Farber disease. Indian Pediatr 2002; 39: 502.
8. McGovern MM, Desnick RJ. Lipidoses. *In*: Behrman RE, Kliegman RM, Jenson HB, Stanton BF. Nelson Textbook of Pediatrics. 18th edition. Philadelphia: WB Saunders Company.p. 593-601.