These bony defects occur over the fontanelles, saggital sutures or inferior angles of parietal bones; this can be explained on the basis of delayed bony union at these sites. At times in the underlying brain development was the cause of the lack of development of the skull bones and that the brain malformation may take the form of local overgrowth of the walls of the neural tube. However, we could not demonstrate any evidence of overgrowth of brain in the CT scan and defect was in the parietal bone itself rather than at angles where the sutures are present. We feel that there must have been a scalp defect which healed *in utero* as evidenced by thick scarred skin at the site of bony defect.

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Farber's Disease

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Farber's disease is a rare genetically determined lysosomal storage disorder of lipid metabolism. Classically it manifests in infancy and consists of a triad of subscutaneous nodules, arthritis and laryngeal involvement. There may be moderate nervous system dysfunction and the lungs, heart and lymph nodes may also be involved. Only 40 cases have been reported so far(1) and to the best of our knowledge, this is the first case report from India.

Case Report

A 3-year-old boy, born of a nonconsan-

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guinous marriage, was brought to the outpatient services of Nehru Hospital, PGIMER, Chandigarh with complaints of delayed and difficult walking and multiple nodules over the joints which appeared at 8 months of age. The swellings were first noticed over the small joints of the hands, then progressed in number and size over the feet, scalp and back. They were described as initially painful erythematous papules evolving into yellow non-tender nodules causing limitation of joint movements. The boy could not use his hands well or walk straight because of restricted joint mobility. The rest of his developmental milestones were reportedly normal. He had recently started having hoarseness of voice. An elder male sibling had died at 8 months of age of an unspecified respiratory illness but had no such nodules. A younger brother aged 3 months, was reportedly normal.

On examination, his anthropometry was normal. He has multiple yellow nodules 4-5 mm in size and 2 to 3 in number over all the interphalangeal joints of the hands and feet with flexion contractures and restriction of movements (*Fig.*). There were moderate flexion contractures at the hip and knee joints as well. Similar nodular swellings were present on the scalp and back. The rest of the symstemic examination was essentially normal. There was no hepatosplenomegaly or lymphadenopathy. There were no neurological findings and the fundi were normal. Fine needle aspiration cytology of the nodules revealed typical 'foamy' cells, with surrounding lymphocytic and plasma cell infiltration forming granulomas characteristic of Farber's disease.

Discussion

Farber's disease, also termed lipogranulomatosis was first described in 1952(2). Presently, the phenotype has been subdivided into 6 subtypes. Type I or the classic

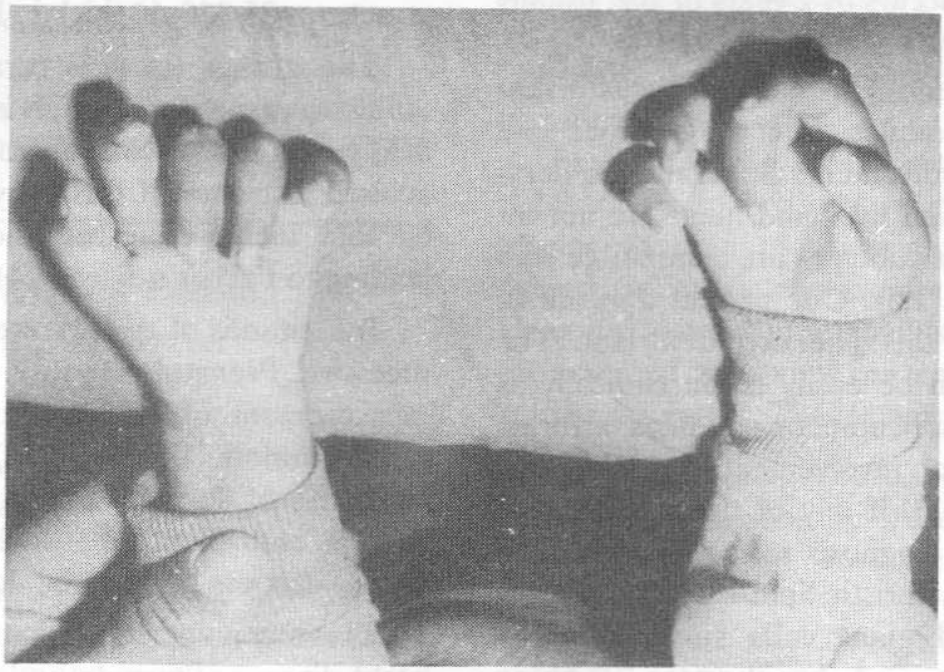

Fig. Multiple nodules and contractures at joints of both hands.

form typically presents in infancy with the appearance of painful joints followed by periarticular swellings and formation of characteristic yellowish nodules in affected areas, especially involving interphalangeal and metacarpophalangeal joints, leading to restricted movements and contractures. The nodules also involve other joints, pressure points, the scalp and other parts of the body. Generalized pigmentation may occur. Lymphadenopathy and hepatomegaly are frequent, splenomegaly is rare. The initial finding in some patients is hoarseness of voice and respiratory obstruction due to involvement of epiglottis and arytenoid folds is commonly reported(3). In addition, these infants have respiratory distress due to pulmonary infiltration and infections.

Neurological impairment may be difficult to assess, but lower motor neuron limb weakness, loss of milestones, and blindness often occur. A cherry-red spot may be seen in the fundus(4). The course of the disease is characterized by repeated chest infections, diarrhea, vomiting and severe weight loss. Death occurs by 2 years of age, usually due to respiratory involvement.

Types 2 and 3 present mainly with subcutaneous nodules, joint deformities and laryngeal involvement. Generally the nervous system and liver and lungs are not involved. These subtypes are compatible with a longer life(1,5). Our patient appears to suffer from this phenotypic variant. The diagnosis can be easily made clinically as the triad of subcutaneous nodules, arthritis and laryngeal involvement is unique for this disease(1). If any of these features is missing, the diagnosis rests on demonstration of characteristic lipid-laden foamy histiocytes and giant cells surrounded by plasma cells and lymphocytes, forming granulomas in the subcutaneous nodules or affected tissues. Bone marrow involvement also occurs. This storage material in PAS-positive and stains with Alcian blue and Sudan Black B(4) . Specific diagnosis may be made by demonstrating, a deficiency of acid ceramidase in cultured skin fibroblasts or in white blood cells. However, this assay is available in only a few laboratories(1).

X-ray chest may show diffuse micronodular pulmonary shadows. Bony involvement leads to osteopenia. Periarticular swellings are prominent(3). There is a non-specific elevation of CSF protein. Associated hypothroidism, and raised ESR and alpha-2 globulin have also been reported(4).

Pathological findings in autopsied cases consist of typical lipid-laden storage cells in all affected tissues. In the CNS there is neuronal storage, with loss of other neurons especially involving anterior horn cells. Characteristic storage material is seen occasionally in neurons of the cerebral cortex and ganglion cells of the retina(4). Electron microscopy reveals curvilinear, tubular and micro-filamentous structures 25-330 A° thick in lysosomes(5).

The storage tissue is ceramide, a normally occurring lipid which is degraded by acid ceramidase into sphingosine and fatty acids. Deficiency of this enzyme causes 5-60 fold increase in ceramide deposition leading to Farber's disease(4).

Inheritance of the disease is autosomal recessive. Prenatal diagnosis is possible by measurement of acid ceramidase in cultured amniotic fluid cells. The enzyme is about 5% of normal value in affected individuals, and about 50% in heterozygotes, so that carriers can also be detected(6).

There is no specific treatment for Farber's disease. Steroids and chlorambucil may improve the joint symptoms(5), but do not affect the ultimate prognosis.

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Normal Values for Penile Standards in Newborns

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Estimation of penile size is important in evaluation of ambiguous genitalia or small genitalia in the genetic males, identification of some syndromes and assessing the effectiveness of testosterone therapy in some conditions. Feldman and Smith have set forth standards for penile size for premature and full term babies of Caucasian origin(1). However, no standards are available for Indian babies. There may be ethnic differences in various morphological and body measurements. In the present study we have tried to set forth norms for penile size for premature and full term infants.

Material Methods

To define standards for penile size in the newborn, 454 full term and preterms (with a range from 26 to 42 weeks) were subjected for measurement of penile size(1). Babies with malformations were excluded from the study. Gestational age was calculated from the first day of the last menstrual period and in every case, clinical assessment of gestational age was preformed by the Dubowitz scoring system(2). All measurements were made between 36 to 48 hours by one of the authors. The data were divided into a series of gestational age group categories. Normal values are presented as mean and ±2SD for different gestational ages.

Penile length was measured from the public ramus to the tip of the glans penis by placing the end of a straight edge ruler against the public ramus applying traction along the length of the phallus to the point of increased resistance, an easily appreciated end point(1). The location of the tip of

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