Seventy per cent develop temperomandibular (TM) joint ankylosis (as in *Case 2)* which used to lead to death by starvation(2). Other common sites of ossification include joint capsules, ligaments and plantar fascia. However, connective tissue of facial and extraocular muscles, intestines, tongue, larynx and skin are not affected. Trauma is the most important exacerbating factor and biopsies should, therefore, be avoided(2). Episodes of ossification are, however, usually spontaneous. Operations in -uninvolved sites are uneventful. Surgical removal of the extra new bone is not helpful as reformation of the myositic bone occurs within a short time. Physiotherapy, change in type of work and genetic counselling should be advised. Numerous drugs have been tried for the primary disease but without success.

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Joubert Syndrome

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Joubert Syndrome is an autosomal recessive anomaly characterized by agenesis of the cerebellar vermis. The symptoms comprise of episodic panting tachypnea, rhythmic protrusion of the tongue, and jerky eye movements in the neonatal period with subsequent ataxia, dysequilibrium, and psychomotor retardation. There are no reports of this condition in the Indian literature. We present a neonate with Joubert Syndrome to alert pediatricians to resort to cranial imaging in neonates with unexplained panting tachypnea.

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Case Report

A boy, the third child of a third degree consanguineous marriage between healthy Indian parents was born by an uncomplicated vaginal delivery at term, after an uneventful pregnancy.

At birth, the infant's weight was 3100 g, length 51 cm and head circumference 38 cm. Apgar scores were 8 at one minute and 9 at five minutes. He was noticed to have a large bulging posterior fontanelle. At 4 hours of age, he was admitted to the Neonatal Intensive Care Unit for respiratory distress, where it was noticed that the breathing pattern consisted of episodes of tachypnea alternating with apnea, bradycardia and cyanosis. The tachypneic episodes lasted upto 30 minutes with respiratory rates of 120-140 per minute. Jerky, irregular conjugate eye movement and generalized hypotonia were present.

Complete blood count, renal and liver function tests, ammonia, lactate, arterial blood gases, and plasma and urinary amino acids were all normal. Bacterial cultures of blood, cerebrospinal fluid, and urine were negative. Chest and skull radiographs were normal. Fundoscopy showed normal discs and maculae.

The abnormal breathing pattern without alteration of arterial blood gases led to the suspicion of Joubcrt Syndrome. An ultrasonography of the skull showed a posterior fossa cyst, mild hydrocephalus and cerebellar hypoplasia. Computerized tomography of the brain showed an enlarged fourth ventricle elevating the tentorium, cerebellar hemisphere hypoplasia and vermis agenesis *(Fig. 1),* thus confirming the clinical diagnosis.

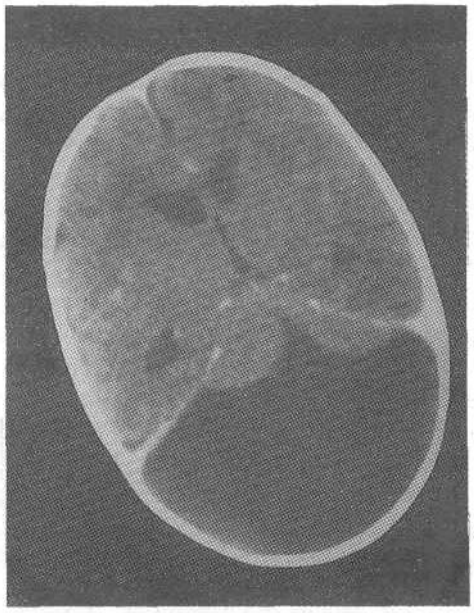

Fig. 1. SCT Scan showing the fourth ventricle communicating with a large cyst m the posterior fossa.

On follow up there was generalized development retardation. At 3 months, the child was unable to control his head, and did not smile or follow objects. Tendon reflexes were normal. Irregular jerky eye movements, a divergent alternating squint, and nystagmus were noted. Fundoscopic examination was normal. The child continued to have episodes of tachypnea without cyanosis or apnea during the waking and sleeping state. Chromosome studies were normal. Ultrasonographic examination of the kidneys showed no abnormality. The child died at 14 weeks of age of acute gastroenteritis.

Physical, neurological, and fundoscopic examinations of both parents and the second sib were normal. However, the first sib (first

of twins), a 3-year-old male was detected to have phenylketonuria, another autosomal recessive disorder.

Discussion

Joubert *et* al.(l) in 1969 recognized the association of developmental defects of the cerebellar vermis with mental retardation, episodic tachypnea and abnormal eye movements in four sibs and one sporadic case. Boltshauser *et al.* (2,3) distinguished it from Dandy Walker syndrome and simple aplasia of the vermis. Subsequently, variable features have been described in Joubert Syndrome including occipital meningoencephalocele, chorioretinal colobomata, dysmorphic facies, hemifacial spasms, polydactyly, renal cysts, seizures, EEG abnormalities, skeletal abnormalities and agenesis of corpus callosum(4,5).

The clinical hallmark of the syndrome is neonatal episodic hyperapnea resembling the 'panting of a dog'(4). However, the abnormal respiratory patterns tend to decrease with age. A detailed neonatal history is thus important in older children.

The ocular abnormalities reported in Joubert Syndrome are retinal colobomata and Lebers amaurosis(4). Lindhout *et al.(6)* in 1980 described a patient with Joubert Syndrome with bilateral chorioretinal coloboma and hypothesized a non-random association. Later, in 1984 Laverda *et al*. (7) reported another patient of Joubert Syndrome with the same ocular defect and Van Dorp *et al.* reported a clinical and pathological description of an affected male and female fetus of the same sibship where coloboma was present(8) further supporting the association. The authors are of the opinion that the association may have been underestimated in earlier reports where electrophysiological studies were not performed. Electroretinography was not done in this patient.

Neuroradiological imaging in Joubert Syndrome shows an unusually shaped fourth ventricle with a midline defect in the posterior fossa-compatible with vermis agenesis. In a clinico-radiological study of 16 cases, CT scan showed dysplasia of inferior vermis which appeared to be split or deeply grooved. There was considerable variation in the size and shape of the fourth ventricle and cisterna magna, but, most commonly, the floor of fourth ventricle was convex towards the brain stem and the superior and inferior cerebellar peduncles were small. Magnetic resonance imaging has revealed an abnormality of brainstem, particularly a small medulla and upper cervical spinal cord(9).

The prognosis in Joubert Syndrome is poor. The 5 year survival rate is 50%(10). There is severe mental and motor retardation. Death is usually due to feeding difficulties and respiratory infections(l0). The recurrence risk for the syndrome is 25%. There is an indication for careful ultrasonography in subsequent pregnancies.

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Jaccoud's Arthritis

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Rheumatic fever is a systemic disease involving joints, heart, pleura, skin and subcutaneous tissue and basal ganglion(l-4). Although, the incidence of rheumatic fever has come down in advanced countries, it still remains **a** common disease in developing countries like India accounting for 30-40% of total heart diseases(l,2). We present here **a** rare manifestation of rheumatic fever Involving small joints of hands leading to **a** prolonged disability and deformity called as Jaccoud's Arthritis(l). Recognition of this entity is important specially for differentiation from JRA (Juvenile Rheumatoid Arthritis) as long-term streptococcal prophylaxis will be required for the former. Only an isolated case report of this entity is reported from India and that too in adults(2).

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Case Report

An 8-year-old child presented to us with persistent joint swelling in ankle, elbow and small joints of hand mainly metacarpophalangeal and proximal interphalangeal joints of one month duration. History of sore throat, palpitation, drug intake, epistaxis,