polyneuropathy with no improvement on gluten-free diet has been reported in two children [2,3]. Cacir, *et al.* [4] reported peripheral axonal polyneuropathy in 2 of the 27 children with CD. Ruggieri, *et al*. [5] reported a girl developing acute demyelinating neuropathy after accidental reintroduction of gluten in her diet, with rapid disappearance of symptoms on a gluten-free regimen. Unlike our case, she had negative serology for CD.

The exact cause of polyneuropathy in CD is not known. The suggested hypotheses include action of antibodies on extra-intestinal tissue transglutaminase proteins [6-9], pathogenic involvement of anti-ganglioside antibodies [8], direct toxic effects of gliadin, and associated vitamin deficiencies (B_6, B_{12} and E). The response to GFD in CD-associated peripheral neuropathy has been inconsistent. Some authors reported symptom regression and resolution of pathological electro-myoneurographic findings on gluten-free diet whereas others reported no benefit of gluten avoidance [10].

In conclusion, an acute polyneuropathy can rarely complicate celiac disease in childhood, and may resolve spontaneously on gluten-free diet.

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Tuberous Sclerosis Presenting with Hemorrhagic Stroke

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Background: Incidence of intracerebral hemorrhage in patients with tuberous sclerosis is rare, and in most of the cases it is associated with either underlying cerebrovascular malformation or hemorrhage into the subependymal giant cell astrocytoma. **Case characteristics:** A 2-year-old boy presented with a hemorrhagic stroke, and subsequently diagnosed as a case of tuberous sclerosis. **Observation:** Detailed work-up for stroke did not reveal any definite etiology. **Outcome:** Weakness gradually improved. Follow-up neuroimaging showed resolution of hemorrhage. **Message:** Clinician must be aware regarding this rare presentation of tuberous sclerosis.

Keywords: *Cerebral hemorrhage, Child, Stroke*.

Tuberous sclerosis complex (TSC) is an autosomal dominant neurocutaneous syndrome with variable penetrance; the estimated frequency is approximately one in 6,000 newborns [1,2]. Although a wide variety of central nervous system abnormalities are associated with TSC, intracerebral hemorrhage is rare [3-9].

CASE REPORT

A 2-year-old boy presented with history of sudden onset repeated vomiting, loss of consciousness, recurrent attacks of generalized tonic-clonic seizures, and left-sided hemiparesis. There was no history of trauma or antecedent surgery, congenital cyanotic heart disease, dehydration, tuberculosis or bleeding disorders. Child was previously admitted to a local hospital where he was treated with phenytoin and aspirin. Past history revealed that he was suffering from recurrent focal seizures since six months of age for which he was prescribed sodium valproate, but the drug compliance was poor for the last three months. He was born out of a non-consanguineous marriage and had two siblings. His elder brother was also suffering from similar kind of focal seizures since early infancy, and had profound mental disability.

At presentation, he was afebrile but comatose with Glasgow Coma Scale of E2V2M3 with stable vital signs, including blood pressure. On examination, he had spasticity and weakness in the left upper and lower limb, positive Babinski sign and exaggerated deep tendon reflexes on the left side. Ophthalmological examination showed mid-dilated pupils with sluggish reaction to light and bilateral papilledema. Child had no signs of meningeal irritation or any cranial nerve involvement. Multiple hypopigmented macules were noted over the face, abdomen and trunk of the child. Other systemic examination was unremarkable.

Routine hematological and biochemical investi-gations were normal. A non-enhanced computed tomography (CT) scan of brain showed massive acute intracranial hemorrhage with perilesional edema located in the right fronto-parietal region with multiple periventricular subependymal calcified nodules (***Fig.* 1a**). MR angiography did not detect any vascular abnormality except intracerebral hemorrhage (***Fig.* 1b**).

Protein C and protein S level, blood lactate levels, Carotid doppler study, and abdominal ultrasound were non-contributory. Ophthalmological examination did not reveal any retinal abnormality. 2D Echocardiography showed two masses, one in left ventricle of size 16×14 mm, attached to the intraventricular septum, and another (10×10 mm) in the right ventricle suggestive of rhabdomyoma. In view of history, clinical examination and investigations, including neuroimaging, the child was diagnosed as a case of tuberous sclerosis. Other family members were assessed; except mother, all had TSC but the clinical presentations were varied. Child improved gradually on medical management and discharged with the advice to continue sodium valproate and physiotherapy. CT scan repeated at 3 month of follow-up visit showed resolution of hemorrhage.

DISCUSSION

Tuber is the characteristic brain lesion of TSC, and may rarely differentiate into a malignant subependymal giant cell astrocytoma. The most common neurologic manifestations are seizures, cognitive impairment, and behavioral abnormalities, including autism [1].

In TSC, intracerebral hemorrhage – with or without intraventricular component – is rare. In most of the reported cases, it was associated with either underlying cerebrovascular malformation, like ectasia, aneurysm and arteriovenous malformation or hemorrhage into the subependymal giant cell astrocytoma [3-9]. The underlying pathophysiologic mechanism of intratumoral hemorrhage remains unclear; increased venous pressure secondary to an increased intracranial pressure is the most accepted pathogenesis leading to necrosis and hemorrhage [6].

In the present case, neither vascular anomaly nor intratumor hemorrhage was detected. Other possible etiologies like hypertension, primary or metastatic central nervous system malignancy, leukemia, coagulopathy, use of drugs like warfarin, amphetamines, cocaine, phenypropanolamine, history of head trauma or antecedent intracranial surgery were excluded. Although the etiology

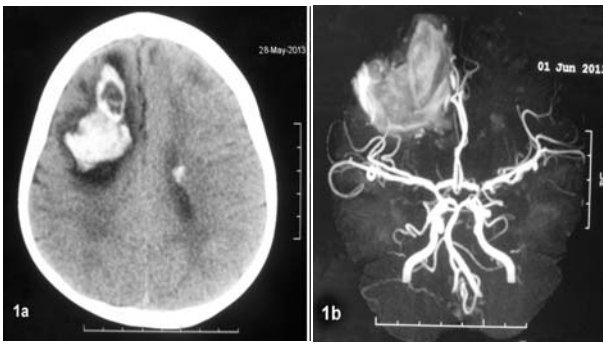

Fig. 1 (*a*) *Non-enhanced CT scan showing large intracranial hemorrhage with perilesional edema located in the right fronto-parietal region as well as multiple periventricular subependymal calcified nodules; (b) MR angiography showing intracranial hemorrhage in the right fronto-parietal region without any abnormality in the major cerebral vessels.*

remains unexplained in the index case, possibility of early hemorrhagic transformation of an underlying embolic ischemic infarction could not be ruled out as the probable pathophysiologic mechanism where cardiac tumours were the potential source of emboli [10].

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Alagille Syndrome with a Previously Undescribed Mutation

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Background: Alagille Syndrome is a rare genetic disease characterized by abnormalities of the intrahepatic biliary ducts with cholestasis along with multisystem anomalies. **Case characteristics**: An 8-year old child with persisting jaundice, severe itching and failure to thrive. **Observation**: Diagnosis of Alagille syndrome was made on the basis of clinical features, typical facies and liver biopsy showing bile duct paucity. Genetic analysis revealed a novel de novo mutation in the JAG 1 gene. **Outcome:** The child was started on ursodeoxycholic acid following which the itching improved. **Message**: A novel *de novo* mutation in JAG 1 gene is described in this child with Alagille Syndrome.

Keywords: *Arteriohepatic dysplasia, Mutation, Posterior embryotoxon.*

Alagille syndrome (AGS) has been classically identified with paucity of bile ducts in the liver, along with involvement of the heart, vertebrae, eyes and typical facial features. AGS has only rarely been described from India [1-4]. We describe a young boy with Alagille syndrome with a previously undescribed mutation in the *JAG1* gene.

CASE REPORT

This 8-year-old boy presented with complaints of persisting jaundice and itching for past 6 years. The parents also complained of the child failing to thrive. For last two years, he was having difficulty in distant vision. He also had history of one episode of blood-tinged vomitus not associated with melena. There was no history of bleeding from any other site or any features of encephalopathy in past. There was no history suggestive of recurrent loose stools or malabsorption. Child was developmentally normal for his age. He was product of non-consanguineous parentage, had two elder siblings