

enzyme assay when such lesions are noted on MRI. Any neuroimaging clue for the diagnosis of neurometabolic disorders are of paramount importance as diagnosis based on clinical information is very difficult and lack of access to biochemical assays in Indian scenario.

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Chiari Malformation Type II with Vanishing Cerebellum

An eight-month-old baby, the second of two siblings born to non-consanguineous parents, presented with delayed milestones and a history of a lumbosacral swelling which was operated in the early neonatal period.

MR imaging revealed a small posterior fossa occupied by the occipital lobes and a profoundly small cerebellum (*Fig 1*). The tentorial incisura was heart-shaped. Sagittal sections demonstrated hypoplastic cord-like cerebellar tonsils herniating through the foramen magnum into the upper cervical canal and an elongated poorly-formed fourth ventricle (*Fig. 2*). Other findings were a small-sized pons with loss of normal pontine

prominence, caudal elongation of the medulla and beaked tectal plate. The torcular heterophili was low-placed and supratentorial hydrocephalus was present. The massa intermedia was absent and the falx was hypoplastic with consequent interdigitations of gyri.

MR images of the lumbosacral spine revealed dysraphism and lumbar meningo-myelocoele with tethering of the cord.

In 1891, Hans Chiari first described an anomaly encompassing elongated peg-like cerebellar tonsils displaced into the upper cervical canal through the foramen magnum to be later known as the Chiari type I malformation. Chiari type II anomaly includes herniation of the medulla, fourth ventricle and cerebellar vermis through the foramen magnum. Chiari III combines features of

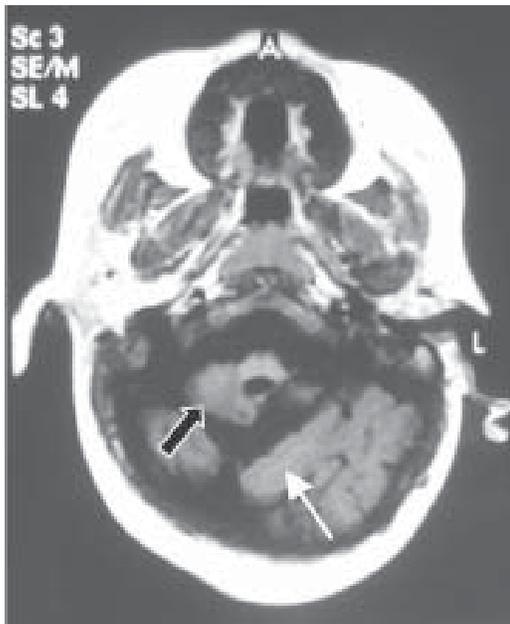


Fig. 1. MRI Axial SE Small posterior fossa occupied by the occipital lobes (white arrow) and a profoundly small cerebellum (black arrow).

Chiari II with a high occipital or low cervical encephalocele. A fourth variety includes severe hypoplasia/aplasia in a diminutive posterior fossa.

The Chiari II malformation is always associated with a meningocele. This condition includes downward displacement of medulla, fourth ventricle and cerebellum into the cervical spinal canal, with concomitant elongated pons and fourth ventricle, probably due to a relatively small posterior fossa.

These intracranial abnormalities are a result of incomplete closure of the neural tube, which prevents transient closure of the central canal that is essential for distension of the primitive ventricular system. The subsequent lack of the inductive effect of pressure and volume on the surrounding mesenchyme results in an abnormally shallow posterior fossa(1). Exceptionally, this transforaminal herniation



Fig. 2. SAG T1 WI: Hypoplastic cord-like cerebellar tonsil herniating through the foramen magnum into the upper cervical canal (Black arrow), elongated poorly formed tube-like fourth ventricle (white arrow), small sized pons with loss of the normal pontine prominence, beaked tectal plate, absent massa intermedia

results in 'degeneration' of cerebellar tissue, presenting as 'the vanishing cerebellum in Chiari II malformation(2). The absence of a normal-sized posterior fossa precludes the diagnosis of cerebellar agenesis(3).

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First-Cousin BMT in Thalassemia with Thymogam Conditioning

Allogeneic bone marrow transplantation (BMT) is the only curative therapy for thalassemia major. The cure rates in class 1 thalassemia are 90-95% and about 70-80% in class 3 thalassemiac children(1). However, only 25-30% of these children have a HLA identical sibling available as a donor for possible BMT(2). In such a situation the parents and first cousins are an option as a donor, when a history of consanguineous marriages is a custom in certain families. We had one such situation where a sibling donor was not available, but a first cousin of the patient was HLA identical, and was hence used as the donor.

The conditioning regimen used for bone marrow transplant in thalassemia involves the usage of ATGAM (Equine antithymocyte globulin). This drug is used as an immunosuppressive agent to suppress the T-lymphocytes. Thymogam is anti-thymocyte globulin harvested from horses immunized with T-lymphocytes and is manufactured in India. Thymogam costs half the price of ATGAM and has been used in the treatment of aplastic anemia in India(3) and also in allogeneic peripheral blood stem cell transplant(4). However it has not been used as conditioning agent in BMT. We recently used thymogam (30 mg/kg/d from day 4 to day 2) for

conditioning regimen in BMT for thalassemia with an intention of reducing costs. The effort was successful and we recommend this conditioning as one option in patients with thalassemia major.

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