A Rare Case Report of Maple Syrup Urine Disease Associated with Dandy Walker Syndrome

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INTRODUCTION

Dandy-Walker malformation is a rare neurological and imaging manifestation [1]. It can affect 1 in 10000 to 30000 newborns [2]. It involves the cerebellar vermis and fourth ventricle [1] and characterized by ventriculomegaly, agenesis of the cerebellar vermis, enlargement of the posterior fossa, hydrocephalus and elevated tentorium cerebelli [3,4]. Maple Syrup Urine Disease (MSUD) is an autosomal recessive metabolic disorder result from defect in the branched-chain α-ketoacid dehydrogenase complex, and contribute to increase the level of the branched-chain amino acids (BCAAs) (leucine, isoleucine, and valine) in plasma, α-ketoacids in urine, and production of the pathognomonic disease marker, alloisoleucine [5]. MSUD is estimated to affect 1 in 185000 infants in the world, frequently the Mennonite population [6]. This can lead to irreversible neurological complications [5]. We here report an unusual presentation of Maple syrup urine disease (MSUD) in an 18-month-old girl presented with Dandy-Walker malformation.

Keywords: Maple syrup urine disease (MSUD), branched-chain amino acids (BCAAs)

CASE REPORT

An 18-month-old baby girl, diagnosed to have Maple syrup urine disease (MSUD), recurrent supracondylar, and right clavicle fractures. She presented to pediatric clinic for increase head circumference, recurrent episode of abnormal movements (probably seizures) and suspicion of blindness (Figure 1).

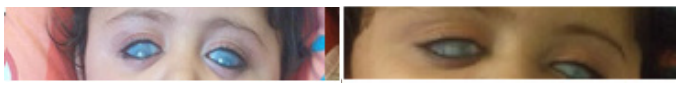
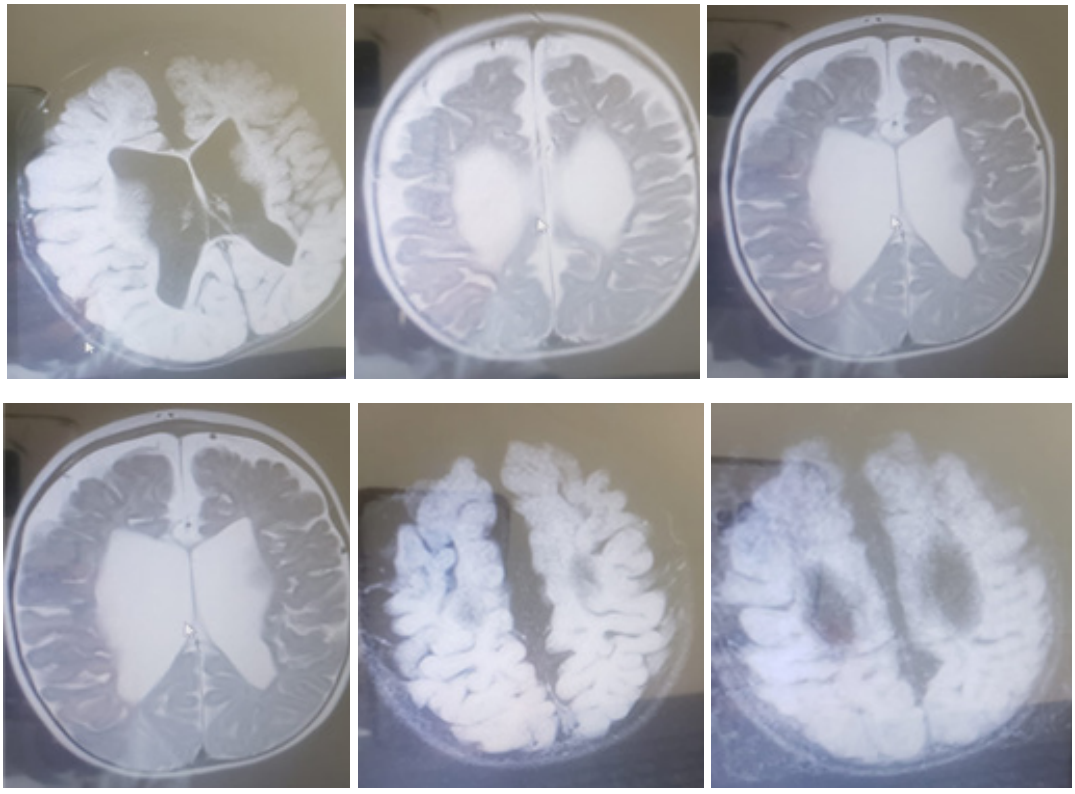


Figure 1: 18-month-old female with blindness

The mother noticed that the girl was floppy since birth, with macrocephaly in addition; she was on antiepileptic drug. As family history: the parents are both in the third decade of life, with no consanguinity. No family history of metabolic disease or any syndrome. They have another normal baby boy. Moreover, the pregnancy and the delivery course were normal, no significant issue. The physical exam showed a hypotonic baby,

with significant macrocephaly. There is a neurodevelopmental delay, poor grimaces or coordination. She had bilateral white eyes. Accordingly, Karyotype done to rule out abnormal numbers or structures of chromosomes to reveal normal; a hypocalcaemia was detected attributed to vitamin D deficiency. An ophthalmologist consultation confirmed that there is a nodular corneal opacity leads to blindness. Finally, MRI brain done showed: cerebellar agenesis, hypoplasia of the corpus collosum, mid brain and severe hydrocephalus, widening of the subarachnoid space and sulci; a diagnosis of Dandy Walker deformity was established (Figure 2).



Figures 2: Magnetic resonance imaging demonstrated absence of the corpus collosum, cerebellar agenesis, enlargement of the ventricles and hydrocephalus: Dandy Walker Deformity.

DISCUSSION

This reported case is patient with MSUD presenting a rare brain imaging with Dandy Walker syndrome. As mentioned before, MSUD is an inborn error of metabolism that can present with several neurological signs and symptoms such as apnea, seizures, and coma as well as chronic features such as poor feeding, ataxia, motor delay, and intellectual disability due to amino acid and neurotransmitter imbalances [7,8]. Its association to neurological malformation is not common. In addition, Dandy Walker malformation is the result of hindbrain maldevelopment [9]. This syndrome can be isolated or be a part of chromosomal abnormality [10], rarely inbron error of metabolism. Furthermore, it can occur in1of 25,000 to 30,000 newborns [11] and some patients are asymptomatic [12]. The literature review of the association of Dandy Walker disease to inborn error of metabolism showed only two cases reported. Firstly, LIU et al reported a very rare association of methylmalonic acidemia and Dandy Walker in a 3-month-old girl with mental retardation [13]. Moreover, in 2012, a patient with non-ketotic hyperglycemia admitted to hospital for myoclonic seizure was diagnosed to have Dandy Walker malformation [14]. These two cases reported Dandy Walker with metabolic disease, but no one reports before MSUD, this can make a very important rare finding. Finally, we can add to our knowledge a new finding: that Dandy Walker can be a neurological manifestation in patients with MSUD.

CONCLUSION

Dandy Walker is rare neurological malformation in patient with metabolic disease, but it can be associated with MSUD as in this case. From this, the importance of MRI brain in all MSUD Patients to eliminate hindbrain malformation.

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