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Case Report

Gluteric aciduria (type-I) Presenting as Bilateral Subdural Hygroma- A Case Report with Literature Review

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Abstract:

Glutaric aciduria type 1 (GA-1) is an autosomal recessive rare neurometabolic disorder caused by absent or deficient activity of glutaryl- CoA dehydrogenase. Patient with gluteric aciduria are more likely to develop collection in the subdural space secondary to cerebral atrophy and expansion of CSF spaces. A 1 year and 10 months toddler presented with increased head circumference since birth and generalized seizure for last few days. Neuroimaging showed bilateral subdural collection of CSF intensity. Bilateral subdural peritoneal shunt was inserted. With early diagnosis and adequate treatment , the majority of children with gluteric aciduria type-I are able to live normal. However treatment for gluteric aciduria type-I must be continued for life to avoid life-threatening complications

Keywords: *Glutaric aciduria, Glutaryl- CoA dehydrogenase, Subdural collection*

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Introduction:

Glutaric aciduria type I (GA-1) is caused by a deficient activity of the enzyme glutaryl-CoA dehydrogenase . It is very rare evidenced by it's prevalence reported 1 in 110000. Deficiency of the enzyme results in accumulation of the gluteric acid and 3-hydroxygluteric acid which has a cytotoxic effect and cerebral atrophy leading to subdural collection . Wide varieties of neurological presentation was recorded in previous studies such as macrocephaly at birth or shortly after, dystonia, and many times resembling seizures at the first episode, with degeneration of the caudate and the putamen. Primary treatment targeting carnitine supplementation to remove glutaric acid, a diet

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restricted in amino acids capable of producing glutaric acid. Treatment of life threatening secondary effects should also be done promptly. With timely dealing of life threatening conditions and certain restrictions for lifelong can make a patient able to live normal.

Case History:

A 1 year and 10 months old male toddler presented with a H/O increased head circumference since birth with significant developmental delay and recurrent episodes of seizures. Complete head control had not been achieved yet. On examination, Occipitofrontal circumference was 53 cm (expected around 47 cm), hypertelorism, global developmental delay. Baseline investigations were within normal range. Spot urine for organic acid showed marked elevation in both gluteric acid and 3-Hydroxygluteric acid levels (Fig:1). MRI of brain revealed large sylvian fissure on both sides with fronto-temporal atrophy, increased signal intensity of the basal ganglia, CSF density accumulation in subdural space on both side (Fig:2).

Surgical description:

Patient undergone bilateral subdural-peritoneal shunt.

Patient was positioned supine. 'C" shape incision was made over parietal region bilaterally. On abdomen linear paramedian incision was made on both side. Cranial end was inserted in to subdural spaces. Abdominal end inserted towards the RIF.

Postoperative imaging showed catheter was in situ (Fig:3)

			Biochemistry	
Test Name	Result	Unit	Reference Range	Method
			ORGANIC ACID URINE	
ORGANIC ACID URINE (SPOT)				
Impression: Marked elevation in both Glutaric acid and 3-Hydroxyglutaric Acid levels				16/03/2
Elevation of both the markers is highly suggestive of Glutaric Aciduria Type-1				11:46AM
Glutaric acid may also be elevated in renal failure and mitochondrial dysfunction				
3-Hydroxyglutaric acid may also be elevated under following conditions				
1.Short Chain 3-, Hydroxyacyl-CoA dehydrogenase deficiency				
2.Severe Ketosis				
Advice:				
1. Plasma acyl carnitine				
2. Enzyme assay for Glutaryl CoA dehydrogenase activity or GCDH gene mutation to confirm diagnosis of Glutaric				
Aciduria Type-1				

Note: Outsourced from Dr Lal PathLabs, India.

Figure 1: *Spot urine for organic acid showing marked elevation in both gluteric acid and 3-Hydroxygluteric acid levels*

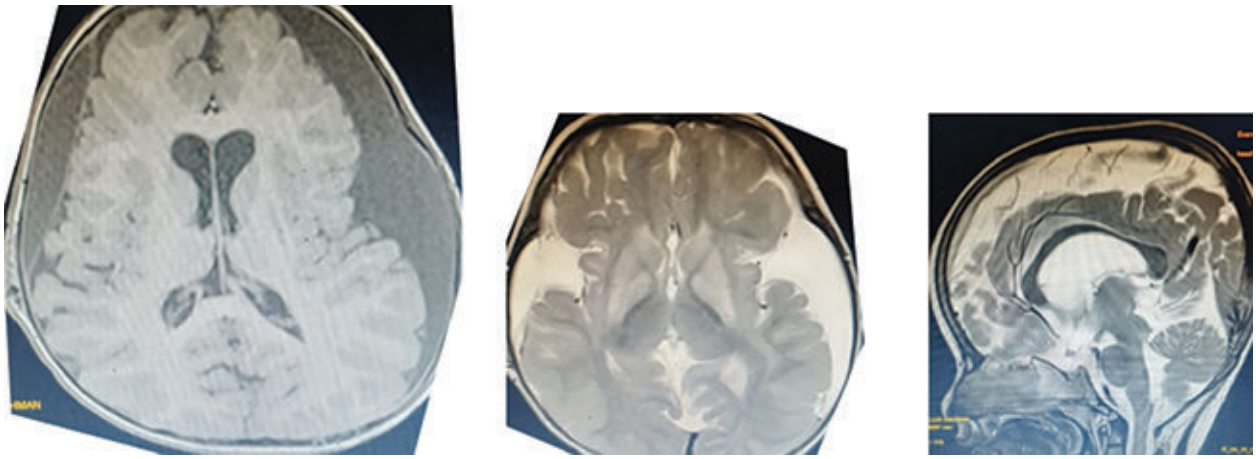

Figure 2: *MRI of brain showing large sylvian fissure on both sides with fronto-temporal atrophy, increased signal intensity of the basal ganglia (T2WI), CSF density accumulation in subdural space on both side*

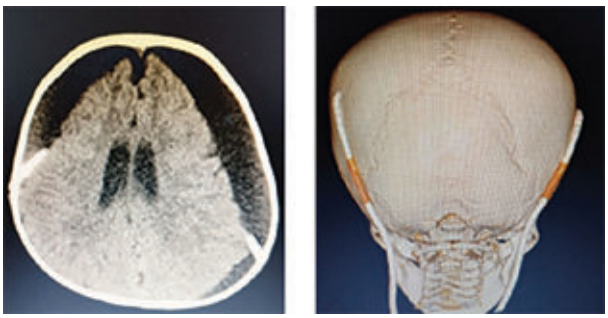

Figure 3: *Postoperative CT scan of brain showing catheter in both subdural space*

Postoperative analysis:
In early postoperative period no episode of seizure was noticed. Symptomatically patient has improved. Concurrent treatment of gluteric aciduria type 1 was started. Follow-up schedule was planned as per recommendation.

Discussion:
Gluteric aciduria type-1 is an autosomal recessive disorder having mutations of the GCDH gene on chromosome 19p13.2 [1]. Macrocephaly is a common feature indicating intracranial pathology [2]. The usual age range presenting for GA-1 is 6 months to 2 years of life [3]. Accumulation of the gluteric acid and 3-hydroxygluteric acid in brain is the pathognomonic for intracranial symptoms which can be measured by urine testing for organic acids [4]. MRI is the investigation of choice having the earliest feature is frontotemporal atrophy [2]. Due to cortical atrophy there is a high chance of subdural collection like hematoma of hygroma [5]. Life threatening conditions should be managed first. Management protocols should include early diagnosis, prevention of acute intracranial episodes like seizure or raised intracranial pressure, with concurrent management of gluteric aciduria in the form of some dietary restriction like low protein diet restricted in lysine and tryptophan, administration of pharmacological doses of riboflavin, and supplementation of L-carnitine form the mainstay of therapy [6].

Conclusion:
Gluteric aciduria type-1 is a rare but treatable disease. As the patient with gluteric aciduria may present with multiple intracranial abnormalities, Timely diagnosis and start of treatment are likely to result in a better outcome. However, even with treatment, 25 to 35% of children with gluteric aciduria type-1 develop some level of motor and intellectual impairment.

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