ISSN: 2689-422X
DOI: 10.33552/GJPNC.2022.04.000580
Global Journal of Pediatrics & Neonatal Care
Iris Publishers

Case Report

Classic Imaging Characteristics of a Clinically Symptomatic Glutaric Aciduria Type I in A female infant- A Case Report

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Received Date: September 09, 2022

Published Date: October 11, 2022

Abstract

This is a case of a female infant who presented with generalised tonic clonic seizures, abnormal movements and fever. She had a background history of failure to thrive and delayed motor development.

Patient demonstrated a classic imaging feature of the Glutaric aciduria type 1, showing widening of the subarachnoid spaces in the frontal lobe and anterior to the sylvian fissures and temporal lobes. There was diffuse T2/FLAIR hyperintensity with corresponding DWI hyperintensity of the deep white matter, lentiform nuclei, caudate nuclei, substantia nigra and dentate nuclei. Her genetic assays showed mutations in the GCDH gene, thus confirming the diagnosis of GA-1.

This report emphasises the need for high index of suspicion in patients with mimics of encephalopathy with preceding febrile illness to have neuroimaging, in order to assess for the possibility of glutaryl aciduria type 1.

Keywords: Glutaric aciduria; GCDH gene; Encephalopathy; Infant

Case Presentation

The patient was an 8month old female infant who presented with poor appetite, fever, and generalized tonic clonic seizures and dyskinesia. There was a background delayed motor development and failure to thrive. On examination she was irritable and mildly dehydrated. She was unable to sit even with support, demonstrated brisk reflexes, spastic muscle tone and dystonia. There was positive consanguinity. The patient was born of healthy parents, and also had a healthy brother. She was a product of normal spontaneous vaginal delivery without any prenatal or perinatal complications.

She had an initial computed tomography (CT) scan at presentation. The showed bilateral widening of subarachnoid space along the frontal convexity, and the space anterior to the temporal poles and sylvian fissures (Figure 1). The patient later had magnetic resonance imaging (MRI) within 24hours of presentation. The MRI confirmed the previous CT findings, and also showed diffuse FLAIR/T2 hyperintensity of the deep white matter, caudate nucleus and lentiform nucleus. Similar signal changes were noted in the substantia nigra and dentate nuclei (Figure 2).

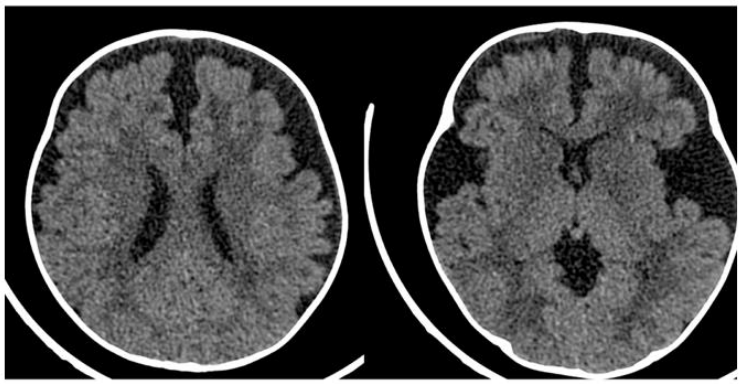

Figure 1: Axial non-contrast CT showing widening of the subarachnoid CSF spaces in the frontal lobes, and anterior to the sylvian fissures in the temporal lobes.

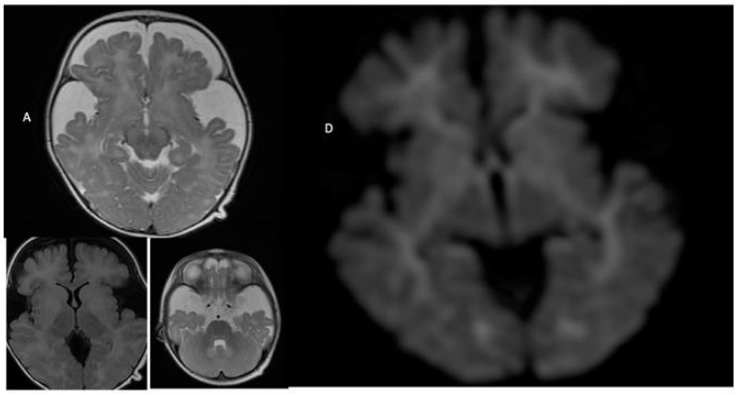

Figure 2: Axial MRI images in T2 (A, C), FLAIR (B) and DWI (D). They show diffuse abnormal hyperintense signal in the deep white matter, lentiform nuclei, caudate nuclei and dentate nuclei. Widening of the subarachnoid CSF spaces and anterior to the sylvian fissures. Corresponding hyperintensity on the DWI image.

She eventually had a genetic assay which identified a heterozygous likely pathogenic variant and heterozygous variant of uncertain significance in the GCDH gene, which were suggestive of autosomal recessive Glutaric aciduria type I.

Discusson

Glutaric aciduria type I (GA-1) is a rare genetic autosomal recessive disorder belonging to a group of cerebral organic acid disorders. There is deficiency of the glutaryl-coenzyme A (CoA) dehydrogenase, with resultant inability to catabolise amino acids: L-lysine, L-tryptophan and L-hydroxylysine. Glutaric acid, 3-hydroxyglutaric acid, and glutarylcarnitine thus become heavily accumulated within the brain and other body fluids [1,2]. Consanguinity is a known risk factor for GA-1 and has a disease prevalence of about 1:100000 newborns [2-4].

Diagnosis is confirmed by localisation of two disease linked GCDH gene mutations or grossly reduced activity of the GCDH in skin fibroblasts or in leucocytes to less than thirty percent [2]. At presentation the affected infants are often hypotonic and irritable, with some abnormal movements. There is usually a preceding febrile illness with clinical signs of encephalopathy, mimicking a viral illness [2], just as demonstrated in the index case. Macrocephaly has been documented as a common presentation or later in the course of the disease [1,2]. This is however not yet seen in this case, probably due to age. GA-1 had been documented to mimic other central nervous system abnormalities, thus high clinical suspicion and neuroimaging is of enormous value to arrive at a diagnosis [5].

MRI has been shown has the mainstay of imaging in the diagnostic management of GA-1 [4]. The index case presented with classic imaging features of GA-1 as previously reported by severe publications. This patient demonstrated bilateral symmetrical diffuse abnormal T2/FLAIR hyperintense signals in the deep white matter, caudate nucleus, lentiform nucleus, substantia nigra and dentate nucleus [6-9]. Mariasavina Severino was however able to note some variations from the index case and other previously reported imaging findings, such as less or non-affectation of the caudate nuclei, and occasional affectation of the thalamus [9].

Andrea Righini et al reported a GA-1 in a female neonate from a consanguineous marriage. An initial 22-week prenatal MRI raised a suspicion of the diagnosis, which was eventually confirmed through a neonatal MRI and laboratory assays. The mother had a prior transabdominal ultrasound which showed a widening of the supra-cerebellar cistern and thus raised the suspicion of more serious intracranial structural abnormality [3]. Widening of the supra-cerebellar cistern was similarly shown in the index case (Figure 3).

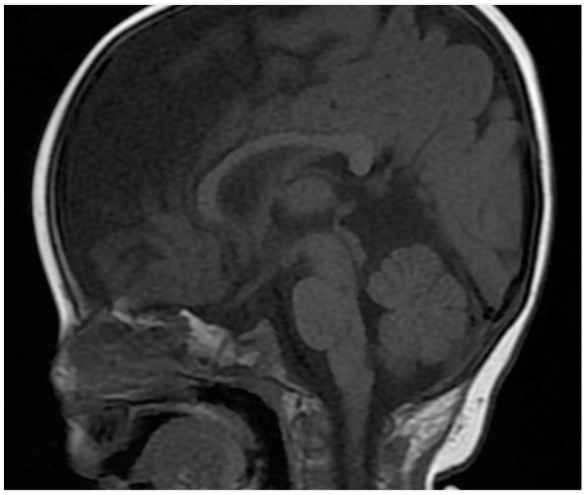

Figure 3: Sagittal T1 image showing widening of the supra-cerebellar cistern.

Conclusion

Despite the rarity of glutaric aciduria type I, the presence of classic clinical signs of encephalopathy preceded by febrile illness, should raise the suspicion of its possibility, and a neuroimaging preferably MRI should be requested.

Funding

None.

Conflicts of Interest/Competing Interests:

None declared by each of the authors.

Consent to Participate

Consent obtained from the parents of the patient, having been assured of complete confidentiality.

Author' Contributions

Each of the authors substantially contributed to the conceptualization, design, acquisition of data and analysis/ interpretation of the acquired patient's data for the purpose of this publication. They all have agreed to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

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