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CASE REPORT

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Propionic acidaemia crisis: a specific DWI finding—case report

Khairy Abdella[1*]

Abstract

Background Propionic acidaemia is among the rarest metabolic disorders described in the literatures. It has resulted from an inborn error in the catabolism of amino acids with an accumulation of organic acids as a consequence of this error. This illness was especially manifested and discovered in early postnatal life with other forms seen later in the childhood period.

It has different clinical presentations with poor feeding, hypotonia, and lethargy described in the early neonatal period, while mental retardation, global developmental delay, and seizures are seen in childhood.

Case presentation We present a case of a 4-month-old child already diagnosed in his early neonatal period with propionic acidaemia currently presented with propionic acidaemic crisis in the form of frequent attack of seizures and severe hyperammonemia with unique DWI features which shows bilateral symmetrical restricted diffusion in the sub-cortical and subinsular white matter that are specific to propionic acidaemia crisis and does not relate to any vascular territory.

Conclusion Propionic acidaemia is one of the rarest metabolic disorders reported in both early neonatal life and the childhood period. Understanding the clinical presentation and MRI findings with specific DWI changes reported in this case assists the health care provider in making the appropriate clinical management and metabolic screening decisions. This case with these rare and specific DWI changes seems to be unique to the propionic acidaemia crisis which reinforces MRI findings and changes mentioned previously in the literatures.

Keywords Propionic acidaemia, Neurometabolicdisorders, Propionyl-CoAcarboxylase deficiency, DWI

Background

Propionic acidaemia is an autosomal recessive disorder [1, 2] which is resulting from an imperfection within the propionyl-CoA carboxylase. Consequently, patients with these disorders exhibit a wide range of metabolic and neurologic abnormalities [2].

Propionyl-CoA carboxylase is responsible for the catabolism of Propionyl-CoA, this error will result in an accumulation of branched amino acids, odd numbers of fatty acids, cholesterol, thymine, and uracil [3].

Propionic acidemia may be displayed clinically early in life or stay to appear afterward in the childhood period with diverse neurological appearances [1].

This report will discuss unique MRI-DWI findings in an infant presented with a propionic academic crisis manifested clinically with severe hyperammonaemia and convulsions.

Case presentation

Already known propionic acidemia 4-month-old child which was diagnosed early in his neonatal period, the patient was on L-carnitine and a low protein diet with periodic routine follow up in the clinic. He was presented

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to the ED with recent attacks of convulsions accompanied with a disturbed level of consciousness.

Upon his arrival and during the clinical examination, he initiated a seizure for 3 min.

Generally, the child was not dehydrated with normal vital signs with oxygen saturation of 95%.

Lab investigation revealed a high level of ammonia 138.2 μmol /L (which is panic), the patient was diagnosed with propionic acidaemic crisis and admitted to PICU, and MRI was requested.

MRI was done during the crisis, DWI sequence (Fig. 1) shows bilateral near symmetrical restricted diffusion involving bilateral subcortical as well as subinsular white matter with both basal ganglia involved as well, confirmed in ADC images (Fig. 2). T2 shows abnormal high signal within the restricted diffusion corresponding areas while T1 and FLAIR show no changes at these areas apart from brain parenchymal hypovolemic changes.

The patient was treated and managed as a propionic acidaemia crisis with follow-up of the ammonia level which shows gradual regression to reach the normal range (45 μmol/L) within the 3rd day after the MRI study.

With no other attack of convulsions recorded and a normal ammonia level as well as a good general condition, the patient was then discharged on his regular follow-up.

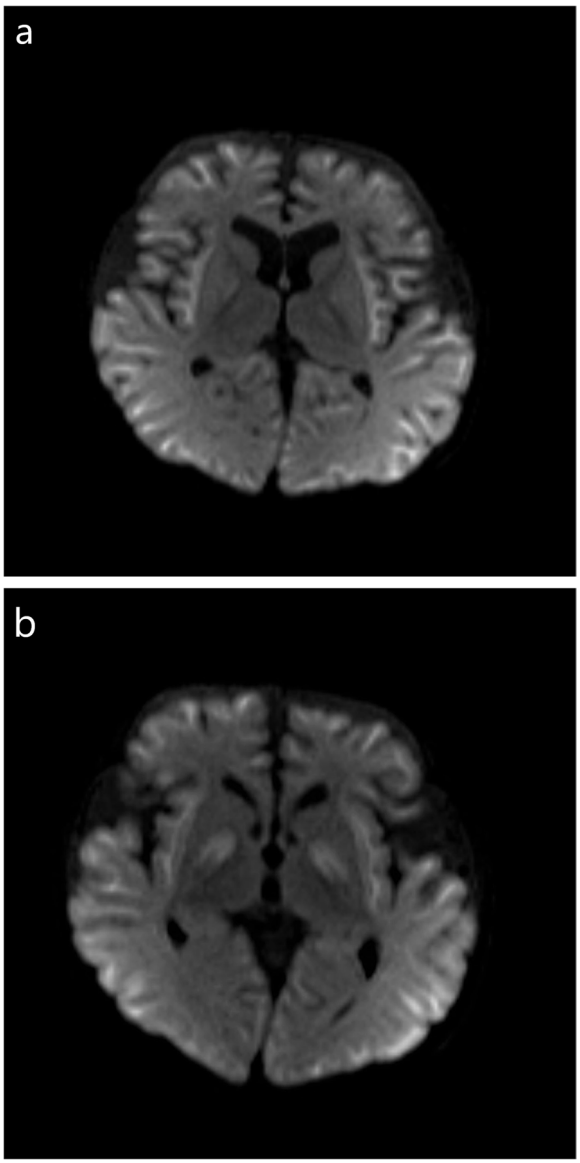

Fig. 1 **a**, **b** Axial diffusion-weighted images shows diffuse bilateral symmetrical restricted diffusion at both subcortical and subinsular white matter and both globi pallidi

Discussion

Propionic acidemia is one of the rarest metabolic disorders affecting the propionyl-CoA (propionyl-CoA) catabolic pathway. In the normal pathway, propionyl-CoA is converted to methylmalonyl-CoA by the mitochondrial enzyme propionyl-CoA carboxylase. Due to this defect, further accumulation of different types of amino acids and different metabolites and subsequent variable neurological manifestations are noted [3].

Incidence of propionic acidemia is varying according to different locations and consanguinity, it is estimated to be 1 in 50,000 to 1 in 500,000 in western countries, where it is noticed to be high in certain areas as Saudi Arabia to be 1 in 2000 to 1 in 5000 and 1:59,426 in Kuwait [4].

The disease manifests itself clinically in a variety of ways, including vomiting, ketosis, hypotonia, difficulty feeding with growth and developmental delay, with convulsions and hyperammonemia which is associated with acute episodes of propionic acidemia crisis [1, 2].

Practice guidelines for the management of propionic acidemia start with dietary control with low protein contents and medication such as L-carnitine, continuous laboratory investigation follow-up, and finally may end in liver transplantation. As a result of the rarity of the disease with subsequent limited peer-reviewed literatures that have been published, the recommendations of these guidelines are rated C or D levels of evidence [1, 2, 5].

Sarah C Grünert, et al. through a cohort study of the impact of propionic acidemia among 55 patients and the clinical outcome, noted that the long-term complications vary between those patients with different neurological manifestations like intellectual impairment, poor IQ with the most pronounced complications, were the hematological one, as pancytopenia and thrombocytopenia and recorded that cardiomyopathy which is

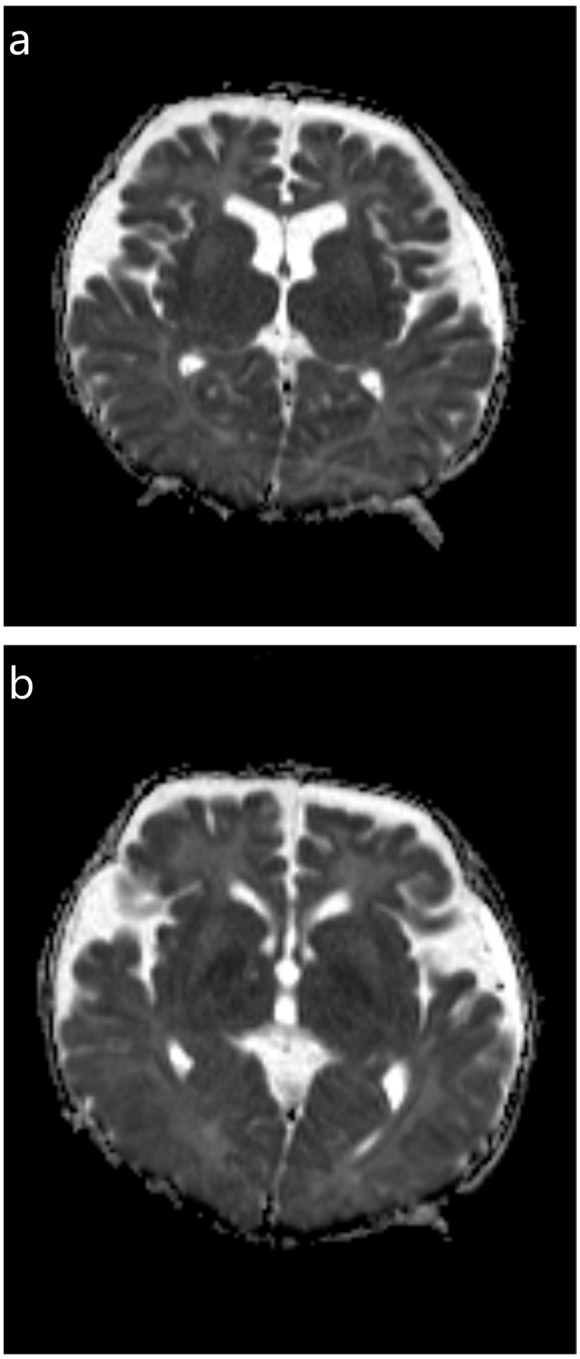

Fig. 2 **a**, **b** ADC images confirm the restricted diffusion at the corresponding areas

associated with high mortality rate is among the serious complications of propionic acidemia [3].

Neuroradiological changes among patients with propionic acidemia are mostly nonspecific and range from white matter changes, basal ganglia affection, and generalized brain atrophy [3].

Cerebral and basal ganglia change pathogenesis is not clear till now with the accumulation of amino acids, especially hyperammonaemia seems to be the most acceptable explanation of these changes [5, 6].

Here we present a case of a rare specific pattern of MRI/DWI findings during the acute propionic crisis with a panic value of hyperammonaemia.

MRI/DWI during the crisis shows restricted diffusion in the subcortical white matter as well as the basal ganglia, this pattern was seldom noted in the literature.

This presentation was found to be unique for propionic acidemia crisis as it is not matched to arterial or venous vascular territory. Also, it is not a typical finding in the case of diffuse hypoxic-ischemic encephalopathy, which tends to be central [7].

Early detection of DWI changes will straightforwardly impact the clinical outcome as well as the therapeutic plans [8].

These changes in DWI are noticeable to be reversible once the crisis has passed and clinical manifestations have returned to normal [2].

Conclusion

Propionic acidemia is one of the rarest metabolic disorders reported in both early neonatal life and childhood period. Understanding the clinical presentation and MRI findings with specific DWI changes reported in this case assist the health care provider in making the appropriate clinical management and metabolic screening decisions. This case with these rare and specific DWI changes seems to be unique for the propionic acidemia crisis which reinforces MRI findings and changes mentioned previously in literatures.

Supplementary Information

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Additional file 1.

Additional file 2.

Additional file 3.

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Author's contributions
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Availability of data and materials
The datasets used and/or analyzed during the current study are available from the corresponding author on reasonable request.

Declarations

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Not applicable.

Consent for publication
Written informed consent for publication of their details was obtained from the patient guardian.

Competing interests
The author declares no competing interests.

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