

# ETHYLMALONIC ACIDURIA AND REPORT OF ONE CASE FROM IRAN

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This condition is due to deficiency in electron transport flavoprotein or electron transport flavoprotein dehydrogenase.

The clinical presentation is characteristic of fatty acid oxidation disorders. This disorder is poor prognostic and death in infancy is common.

The central nervous system involving causes neurodevelopmental delay, hypotonia, and head lag.

Different type of seizures such as infantile spasm and generalized tonic clonic seizure begin in infancy and become refractory to antiepileptic drugs. Also episodes of status epilepticus are frequent.

Neurological exam shows manifestation of upper motor neuron disease including exaggerated deep tendon reflex, ankle clonus and positive Babinski sign.

Infectious disease and Intercurrent illnesses may lead to neurological deterioration and coma or death in the first few years of life. Also hemorrhagic episodes, for example, petechiae, ecchymose, hematuria and blood in stool associated with infectious disease is common manifestation.

The hematologic investigations show no evidence of abnormalities in clotting and platelet function.

The facial features of these patients resemble to each other and are mildly dismorphic. Sometimes the nasal bridge is depressed.

Neuroimaging evaluation revealed delayed myelination and frontotemporal atrophy with high T2 intensity in basal ganglia.

Acute catabolic state with crises of lactic academia and hypoglycemia are frequent but lactate and pyruvate can remain high between attacks.

During attacks lactate level as high as 17 mmol/l and acidosis is more severe with PH values of 7.05 to 7.10.

The major metabolic abnormality is high excretion of ethylmalonic acid in the urine.

This disorder is transmitted in an autosomal recessive trait and boys and girls have been reported in same family.

Ethyl malonicaciduria is lethal during infancy or first few years of life. Treatment with carnitine, vitamin C, vitamin D and riboflavin did not show dramatic effect.

Diet with restricted methionine is helpful for decrease excretion of ethylmalonic acid in the urine and decrease the level of serum lactate and pyruvate.

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### **Case presentation**

A 2-year-old boy was referred to author' clinic for evaluation of neurodevelopmental delay.

He was the product of first pregnancy of consanguineous parents born by cesarean section. He had rolling and creeping but did not have the ability of sitting, standing and walking. He could not speak.

He had a history of admission for bloody stool after 20 days of birth but hematologic evaluation did not confirm abnormal evidence.

Neurological exam showed cerebral hypotonia (hypotonicity with no weight bearing and exaggerated deep tendon reflex). MRI revealed abnormal signal intensity in periventricular white matter and basal ganglia.

Routine lab exam and venous blood Gas, ammonia and high performance liquid chromatography were in normal limits. Serum lactate level was mildly elevated. Urine organic acid showed high ethylmalonic acid 2125 mmol/mol creatinine (normal <17), therefore the diagnosis of ethyl malonic aciduria was confirmed.

**Keywords:** Ethylmalonic aciduria; neurodevelopmental delay; report of a case.