DIAGNOSIS OF ORGANIC ACIDEMIA

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Organic acid occur as physiologic intermediates in variety of intracellular metabolic pathways, such as catabolism of aminoacid, mitochondrial β oxidation of fatty acids, tricarboxilic acid cycle, and cholestrol and fatty acid biosynthesis. The classical organic aciduria represent the pursuit of abnormalities of aminoacid degradation beyond deamination Their diagnostic hallmark is an accumulation of characteristic organic acids.The clinical features result from toxicity of the accumulating methabolites.

Treatment involved 1. protein restriction 2. supplementation of aminoacids with unimpaired metabolism as well as trace elements and 3. specific measures for detoxification if indicated. Diagnostic tests consist of CBC, FBS, Bun, Cr, uric organic acid, TG, Cholestrol Ca, P, ALP, VBG, Na, K, Cl, U/A(PH, SG, Ketone), Ammonia, lactate, pyrovate, Ketone body CPK, Aldolase, SGOT, SGPT, BIL, PT, PTT, Plasma aminoacid HPLC, Homocysteine, Urine aminoacid and carbohydrate chromatography,

Acyl carnitine profile, urine organic acids and for next steps tissue specimen and enzyme activity and gene study.clinical chemical indices of organid aciduria is Metabolic acidosis, Increased anion gap, Hyperglycemia and hypoglycemia, Ketosis and Ketonuria, Lactic acidosis, Hyperammonemia, Hyperuricemia, Hypertriglyceridemia, increase of transaminase Granulocytopenia, thrombocytopenia and Anemia. Acylcarnitine profile and urine organic acids are two for important tests for differentiation of types oforganic academia. Diagnostic profile for diagnosis of organic academia are summarized in following table:

Keywords: Organic academia; plasma acylcarnitine profile; diagnosis.

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Table: Diagnosic profile for organic acidemia

MSUD	P: leucine, valin, isoleucine, alloisoleucine ↑ U: branched chain oxo and hydroxyacids ↑
IVA	U:isovelerylglycine, 3 hydroxyisovaleric acid ↑ U,P,S:isovalerylcarnitine↑ C0:↓
3MCC	U: 3methylglycine,3hydroyisovaleric acid ↑ U,P,S : 3 OH isovalerylcarnitine ↑ C0:↓
MGA 1	U: 3methylglutaconic acid, 3methylglutaric acid, 3 hydroxyisovaleric acid ↑ P,S,U:3 hydroxyglutaconylcarnitine↑
MGA II	U: 3methylglutaconic acid, 3methylglutaric acid, 2 ethylhydracrylic acid ↑
MGA III	U:3methylglutaconic acid, 3methylglutaric acid ↑
MGA IV	U: 3methylglutaconic acid, 3methylglutaric acid ↑
MHBD	U: 2 methyl 3 hydroxybutyric acid, ethylhydracrylic acid, tiglylglycine ↑ U,S,P: C5:1carnitine↑, 2methylacetoacetate: Normal
PA	U: propionic acid, 3 hydroxy propionic acid,propionylglycine ,methycitric acid ↑ P,S,U: glycin,C3 carnitine ↑ P,U: lactate ↑ P,S: C0 ↓ B: NH3 ↑
MMA	U: methylmalonic acid,propionic acid 3 hydroxy propionic acid ,propionylglycin methycitric acid ↑ P,S,U: glycin,C3 carnitine ↑ P,U: lactate ↑ B: NH3 ↑ P,S: C0 ↓
BTD	U,P,CSF:lactic acid ↑ P: C3 ↑ P,S: C0 ↓ U: 3 hydroxy propionic acid, propionylglycin ,methycitric acid, 3methylcrotonylglycine, 3hydroxyisovaleric acid↑ P,S,U: 3 hydroxyisovalerylcarnitine ↑, Biotinidase↓ (diagnostic)
HLCSD	U,P: lactic acid ↑ P: C3 ↑ P,S: C0 ↓ U: 3 hydroxy propionic acid, propionylglycin , methycitric acid,3methylcrotonylglycine, 3hydroxyisovaleric acid↑ P,S,U: 3 hydroxyisovalerylcarnitine↑ Normal Biotinidase
GA I	U: glutaric acid, 3 hydroxyglutaric acid Glutaconic acid ↑ P,S,U: glutarylcarnitine ↑ P,S : C0 ↓
D2HA	U: D2 hydroxyglutaric acid krebs cycle intermediates ↑ CSF: GABA, protein ↑
L2HGA	U: L2hydroxyglutaric acid ↑ CSF,P,U : lysine ↑
ASPA	P,S,U: N acetylaspartic acid ↑
EE	U: ethylmalonic acid, mrthylsuccinic acid,C4-C5 acylglycines ↑ P,S, U: C5, C6 acylcarnitine ↑

P: Plasma, B:Blood, U:Urine, S:Serum, MSUD: Maple urine syrup Idisease, IVA: isovaleric academia, 3MCC:3 methylcrotonyl-CoA dehydrogenase def, MGA1:3 methylglutaconyl CoA hydratase def, MGA II :Barth syn, MGA III:Costeff optic atrophy, MGA IV:methyl glutaconic aciduria IV, MHBD: 2 methyl 3 hydroxybutyryl- CoA dehydrogenase def, PA : proppionyl-CoA carboxilase, MMA: methylmalonyl-CoA mutase def, BTD:biotinidase def, HLCSD:holocarboxilase synthase def, GAI:glutaryl CoA dehydrogenase def, D2HA:D2 hydroxyglutaric acid dehydrogenase def, L2HGA: FAD dep, L2 hydroxyglutarate, dehydrogenase def, ASPA : aspartoacylase, aminoaylase II EE: mitochondrial matrix protein