

Case Report

Glutaric Aciduria Type II Multiple Acyl-CoA Dehydrogenation Deficiencies

* Nabil Barqouni

Introduction:

Glutaric Aciduria type II, is an inborn error of metabolism, autosomal recessive due to defects of mitochondrial electron transport chain from dehydrogenation reaction catalyzed by MCAD, SCAD, LCAD and VLCAD as well as Glutaryl-CoA Dehydrogenase.

These deficiency produce illness in newborn period characterized by acidosis, hypoglycaemia, coma, hypotonia and cardiomyopathy

Some affected neonates have had facial dysmorphic features, polycystic kidneys.

Diagnosis:

Can be made from urinary organic acid fatty acid oxidation block (Ethylmalonate and dicarboxylic acid) lysine (glutarate) and branch-chain amino acids

Most severely affected infants have not survived the neonatal period Secondary carnitine deficiency is present.

Case Report:

Three weeks old female neonate, product of full term, NSVD, in private clinic B.Wt 2.8 kg of multigravida mother and consanguineous parents.

She was admitted in SCBU in EGH with intractable diarrhea, perianal skin peeling, no fever, no vomiting. Clinical examination, she was dehydrated, acidotic breathing, losing wt 2.4kg perianal macerated dermatitis.

Family history revealed two infants were expired with similar problem (boy, girl) at (4-7 months) but no medical report or final diagnosis

Baby was suspected with Inborn Error Of Metabolism (IEOM)

Initial lab investigation showed metabolic acidosis, hypoglycemia, normal ammonia, blood culture was negative.

Blood urine for Amino acid, organic acid chromatography were done and blood zinc level, and skin culture fibroblast.

Investigation were revealed Organic acids profile showed raised levels of Ethylmalonic acid 507mmol/mol crn (N<14.6), Methylsuccinic acid 40, (N<8.8), Malic 361, (N<38), Glutaric acid 46, (N<5.3)

2 Hydroxyglutaric acid 110, (N<69.5), Adipic acid 102, (N<34),

Fumaric acid 93, (N<14).

Skin culture fibroblast confirmed diagnosis.

These result indicates metabolic disease of mitochondrial disorder, glutaric aciduria type II or SCAD.

Baby was given in SCBU, I.V. Fluids, antibiotic, bicarbonate, L-carnitine, Multivitamin.

The baby was improving and discharge from SCBU and follow up in OPD clinic.

At 3 months old admitted with generalized oedema, oliguria, respiratory distress.

Clinical examination revealed severe anasarca, ascites, admitted in PICU

Investigation revealed hypoalbuminemia and gross proteinuria, hypercholesterolaemia, hypocalcaemia. Infant treated as congenital nephrotic syndrome with methylprednisolone, I.V. human albumin, mechanical ventilation, captopril, diuretics but patient deteriorated and expired within 10 days.

Discussion:

Inborn error of metabolism IEOM is an autosomal recessive was noted increasing in Palestinian population most likely due to consanguinity which increased in rural areas especially north and south area in Gaza.

The incidence of IEOM is very rare especially glutaric Aciduria type II, this case was submitted due to rare disease and diagnosed was confirmed by blood and urine and organic acid chromatography and skin culture fibroblast were done in Sheba Medical center, our patient also had congenital nephrotic syndrome most likely (Finish) type, due to some genetic disease and IEOM Case was given treatment mostly supportive and coenzyme, Carnitin, multivitamin riboflavin, Methylprednisolone, captopril but unfortunately most of these patients were not survived.

References:

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MBBCH (Ain shams university)
DCH (RCPI) Dublin
DCH (RCPE) Edinburgh
Jordanian + Arab and Saudi board in paediatric