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LETTER TO EDITOR (VIEWERS CHOICE)

MEDIUM CHAIN ACYL COA DEHYDROGENASE (MCAD) DEFICIENCY IN AN INDIAN NEONATE

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Medium Chain Acyl-CoA Dehydrogenase (MCAD) deficiency, the commonest mitochondrial fatty acid oxidation disorder with an autosomal recessive mode of inheritance and a leading cause of unexplained death is rarely reported in Asian/Indian population.[1,2,3] MCAD deficiency occurs due to mutations in the medium-chain acyl-CoA dehydrogenase (ACADM) gene, which is located ona chromosome 1p31 and comprise of 12 exons spanning 44 Kb.[4] We report probably the first case of MCAD deficiency in an Indian neonate.

A 2 days old male baby born by 2nd degree consanguineous marriage to a non-diabetic mother presented to the neonatal intensive care unit (NICU) after 24 hours of life with refusal to feeds, dull activity and abnormal movements of limbs. He was born at term by normal vaginal delivery, cried immediately after birth and had a birth weight of 3.9 kg. On 30 hours of life, he had cyanosis (room air oxygen saturation (SpO2) of 80%), heart rate (HR) of 86/min, respiratory rate (RR) of 20/min and blood sugar of 36 mg/dl. He was immediately given intravenous (IV) glucose and simultaneously bag-mask ventilation and subsequently intubated and referred to us. On arrival to our NICU, the neonate had RR of 78/min, HR 39/min, capillary refill time (CRT) of 5 secs, blood pressure (BP) of 63/34 mm of Hg with mean arterial pressure (MAP) of 43 and SpO2 of 60% with poor cry/tone/activity. Systemic examination was normal. Baby was resuscitated and required mechanical ventilation for 5 days along with inotropes. Initial ECG showed wide QRS complex with tall T waves and simultaneous serum potassium levels of 7.3 mmol/l. Baby was treated with IV calcium gluconate, glucose insulin infusion, potassium free fluids and per-rectal calcium polystyrene sulphonate powder. Initial hypoglycemia (blood sugar 30 mg/dl) was corrected with IV glucose bolus with and subsequently with fluid requiring glucose infusion rate (GIR) of 10 mg/kg/min for maintenance of euglycemia. Feeds were started at 6th day of life. On further investigation, echocardiography revealed biventricular dysfunction with moderate pulmonary arterial hypertension. Arterial blood gas (ABG) showed metabolic acidosis (pH-7.19, bicarbonate 13 mmol/l). Urine for ketone bodies was negative and serum lactate was 5.1 mmol/L (Normal: 0.5-1.6 mmol/l). Blood and endotracheal secretions culture grew *Klebsiella pneumoniae.* Cerebrospinal fluid (CSF) examination was normal. He was treated with cefotaxime and later shifted to piperacillin-tazobactam for 10 days based on drug sensitivity report. Tandem mass spectrometry (TMS) revealed elevated levels of medium chain acylcarnitines [hexonoylcarnitine (C6) and octanylcarnitine (C8)] and significant elevated ratio of C8/C10 (7.17) which is specific to MCAD. TMS was repeated again on 15th day of life which revealed the same findings. Thus diagnosis of MCAD deficiency was made. Supportive therapy (increased frequency and volume of feeds with added sugar and glucose monitoring at home by glucometer) was given to prevent hypoglycemia and he was discharged on day 23 of life after explaining the need for frequent feeding, avoidance of starvation, monitoring of blood sugar, and to avoid food having medium chain fatty acid. Currently the child is 6 months old and found to have near normal neurodevelopment. Genetic evaluation could not be performed due to non-affordability and attenders have been counseled the need of it in near future.

Exact incidence and prevalence of MCAD deficiency in Indian subcontinent remains unknown.2 When undiagnosed in neonatal period, affected patients generally manifest within 3 months – 5 years with episodes of acute illness precipitated by prolonged fasting (> 12 -16 hrs). Lethargy, vomiting, hypoglycemia, seizure, encephalopathy, hypoketonemia, are usual presentations but can range from asymptomatic to idiopathic hypoglycemia to sudden unexplained death.[2,3] Clinically MCAD was suspected over sepsis in our patient in view of acute catastrophic presentation requiring extensive resuscitation in term healthy neonate at 30 hours of life with no perinatal risk factors for sepsis. Also the child had persistent hypoglycemia and TMS twice was suggestive of MCAD. The second TMS was done when the child was infection free. If MCAD is missed, approximatively 25% of patients die during the initial metabolic crisis or have irreversible neurologic impairment.[2] It is usually diagnosed by TMS during newborn screening program. Mainstay of treatment is avoidance of fasting for more than 12 hours and frequent feeding. Catabolic stress should be promptly treated by restricting dietary fat. There is no clear cut recommendation for carnitine supplementation. The

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prognosis is excellent for those who survive metabolic crisis period without brain damage.

Compliance with Ethical Standards

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References :

1. Ensenauer R, Winters JL, Parton PA, Kronn DF, Kim JW, Matern D, et al. Genotypic differences of MCAD deficiency in the Asian population: novel genotype and clinical symptoms preceding newborn screening notification. Genet Med. 2005;7:339-43.
2. Grosse SD, Khoury MJ, Greene CL, Crider KS, Pollitt RJ. The epidemiology of medium chain acyl-CoA dehydrogenase deficiency: An update. Genet Med. 2006;8:205-212
3. Oerton J, Khalid JM, Besley G, Dalton RN, Downing M, Green A, et al. Newborn screening for medium chain acyl-CoA dehydrogenase deficiency in England: prevalence, predictive value and test validity based on 1.5 million screened babies. J Med Screen. 2011;18:173-181
4. Zhang ZF, Kelly DP, Kim JJ, Zhou YQ, Ogden ML, Whelan AJ, Strauss AW. Structural organization and regulatory regions of the human medium-chain acyl-CoA dehydrogenase gene. Biochemistry. 1992;31:81–89.