Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com/journal/sjmcr/home

OPEN ACCESS

Very-Long-Chain Acyl-Coenzyme a (CoA) Dehydrogenase Deficiency in Saudi Neonate- Case Report

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| **Received:** 10.01.2019 | **Accepted:** 20.01.2019 | **Published:** 30.01.2019
DOI: 10.36347/sjmcr.2019.v07i01.027

Abstract **Case Report**

Very-long-chain acyl-coenzyme A (CoA) dehydrogenase deficiency is an autosomal recessive disorder of fatty acid B-oxidation with significant variable presentation. The Main presentation are hypoketotic hypoglycemic, hepatomegaly, cardiomyopathy, myopathy and rhabdomyolysis, we present a case report of 2 days old Saudi neonate who presented with non ketotic hypoglycemia, hepatomegaly, high lactic acid level and high creatine kinase. He was diagnosed as very long chain acyl coenzyme A dehydrogenase deficiency based on newborn metabolic screen
Keyword: Very-long-chain acyl-coenzyme A (CoA) dehydrogenase deficiency fatty acid B-oxidation.

INTRODUCTION

Very-long-chain acyl-coenzyme A (CoA) dehydrogenase deficiency is an autosomal recessive disorder of fatty acid B-oxidation with significant variable presentation [1, 2]. It is classified into three phenotypes, i.e., early infantile-onset type, childhood-onset hypoglycemic type, and adolescent/adult-onset myopathic type [3]. The most common symptoms are hypoketotic hypoglycemic, hepatomegaly, cardiomyopathy, myopathy, rhabdomyolysis, high creatinine kinase and lipid infiltration of muscle and liver [4].

CASE REPORT

A male infant of healthy consanguineous parents was born at 38 weeks by normal spontaneous vertex delivery to a 25 year old primigravida mother. Pregnancy and delivery were uncomplicated. Apgar score was 9 at the first mint and 10 at fifth mint. At age of 33 hours, infant suddenly presented with tachypnea, subcostal retraction and desaturation down to 75% with bluish discoloration of the face. The infant was admitted to the intensive care unit for sepsis evaluation. He was put on nasal blended oxygen, kept NPO and placed on antibiotic. On the 3rd day of life 4, infant became well, maintain oxygen saturation on room air and orogastric tube feed started. On the 5th day of life infant became tachypneic, desaturated with low random blood sugar (11 mg/dl). Laboratories collected found ammonia 51 umol/L(64-107 umol/L) ,lactic acid level 3.4mmol/l (0.5-2.2 mmol/l), low glucose concentrations 0.1 mmol/l (4.1-5.9 mmol/l) and high creatine kinase 2,263 U/L (39-308 U/L) indicating rhabdomyolysis. Blood urea nitrogen and creatinine were also within normal range while liver function tests revealed mildly elevated transaminases. Urine ketone was negative. Insulin and C peptide were negative during hypoglycemia. Cortisol and ACTH were normal as well. Newborn screen done showed significantly elevated C14:1(2.494) suggesting Very long chain acyl coenzyme A dehydrogenase. Urine organic acid profile showed mild elevation of dicarboxylic aciduria (adipate, suberate and sebacate) and elevated 3-Hydroxysebacic acid and octenedioate. Echocardiography done showed Patent foramen ovale, Patent ductus arteriosus and right ventricular hypertrophy. Ultrasound of abdomen showed hepatomegaly. The infant was started on aggressive hydration with 10% dextrose-containing fluids as well as a medical formula low in long chain fat and enriched with medium chain triglycerides. The creatine kinase declined over the next 3 days reach 323 U/L.

DISCUSSION

Very-long-chain acyl-coenzyme a dehydrogenase (VLCAD) deficiency is characterized by the impaired mitochondria β-oxidation of long chain fatty acids. Clinical symptoms are often triggered by prolonged fasting, physical exercise, or infections.

VLCAD deficiency is clinically classified as follows: 1) a severe form with a high mortality rate exhibiting cardiomyopathy, hypoglycemia, and

hepatomegaly in the neonatal period. Our patient presented with non ketotic hypoglycemia, hepatomegaly, and rhabdomyolysis. 2)A milder form, mainly presenting as hypoketotic hypoglycemia in childhood, without cardiomyopathy. 3) A myopathic form presenting after childhood with intermittent episodes of rhabdomyolysis and myalgia triggered by exercise or fasting [4]. Fatty acid oxidation defects (FAODs) responsible for 5% of all sudden and unexpected deaths in infants [5].

Once the diagnosis of VLCADD is established, symptomatic treatment as well as VLCADD-specific treatment should be administered. Patients should be fed a low-fat formula with MCTs for calorie supplementation. Furthermore, intensive monitoring of cardiac, muscular and hepatic function will ensure a good prognosis. Additionally, it is crucial to avoid conditions such as fasting, dehydration, high-fat diet and myocardial irritation. Intravenous glucose injection to correct hypoglycemia, close monitoring of cardiac rhythm and supportive care for rhabdomyolysis should be performed [6].

CONCLUSION

(VLCAD) is rare neonatal entity and should be considered in neonate with non ketotic hypoglycemia, hepatomegaly and elevated cretinin kinase. High index of suspicion, early diagnosis and management save patient from late complication.

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