Managing a rare deficiency beyond the genetics in immigrant populations: addressing the cultural and social barriers to accessing care

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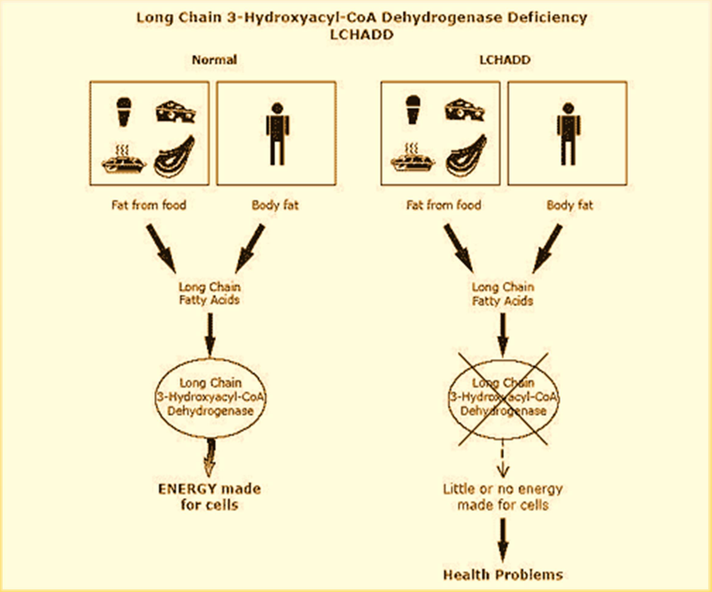
Introduction

LCHAD (Long-chain 3-hydroxyacyl-CoA dehydrogenase) deficiency is an inborn error of fatty acid metabolism caused by a rare autosomal recessive mutation. This enzyme deficiency leads to accumulation of fatty acids which results in symptoms such as hypoglycemia, developmental regression, cardiomyopathy, hepatomegaly, muscular hypotonia, pigmentary retinopathy, and sudden death. Because of the complexity and fragility of this condition, it is crucial that patients receive access to multi-disciplinary specialty care to ensure appropriate management of the severe complications that can occur.



http://adamslchad.com/lchad.html

Case Presentation

Patient is a 2-year-old Peruvian female with a complex underlying metabolic deficiency that is brought to the clinic for her well child visit.

Past Medical History

Patient was born via C/S at 37 weeks to a healthy G1P0 mother whose pregnancy was complicated by hyperemesis and preeclampsia**.** At 8 months of age, developmental regression occurred after prolonged hospitalization. This led to:

- General: Temperature instability and diminished interactivity
- Language regressed from 50 words to 5 words and inability to put 2-3 words together
- GI: Swallowing difficulties leading to gastric tube placement in abdomen.
- MSK: Mild Hypotonia and weakness in lower extremities. She sat at 6 mo. and regressed after hospitalization and only sat again at 12 mo.
- Neurological: intermittent seizure activity and labile emotions with frequent temper tantrums

Course History of Disease

October 2015 (8 mo.)
Episode of hypoglycemic coma with seizure. Patient is clinically diagnosed with LCHAD in native country. Pediatrician refers to genetics department in Miami.

March 2016 (14 mo.)
Patient develops 5th episode of rhabdomyolysis with a CPK value of 16,000. Patient was hospitalized for 2.5 months.

April 2016 (15 mo.)
Patient and mother visit the U.S. Seen by genetics department in Miami.

May 2016 (16 mo.)
Diagnosis confirmed by genetics department (heterozygous splice variant of the HADHA gene).

June 2016 (17 mo.)
Presented to the ER with emesis and difficulty feeding. Patient was hospitalized for 10 days. Gastric tube placed during this visit.

October 2017 (33 mo.)
Reestablishing care with genetics.

November 2017 (34 mo.)
Seen in pediatric mobile clinic for primary care visit.

Discussion

Managing complex metabolic deficiencies like LCHAD requires extensive workup by specialists as well as supportive care due to the many complications that can arise from the deficiency. The referral list for this patient includes but is not limited to cardiology, gastroenterology, endocrinology, neurology, nutrition, and speech therapy. There were multiple social and cultural challenges that prevented the patient from getting regular access to specialty care. As a newly arrived immigrant, she had no fixed residence, her mother did not speak English, no family support network, and no health insurance. The pediatric mobile clinic served an instrumental role in empowering the patient's mother to gain access to the complex US healthcare system by addressing these unique challenges.

The patient's mother was counseled by the clinic's social worker who helped acquire a health center card for the patient granting her access to specialty care. The patient was also encouraged to continue with the WIC program which provided specialized formula to ensure the child received appropriate nutritional support. Patient was provided with education and anticipatory guidance including recognizing the signs of acute distress due to metabolic crisis and when to take the child to the hospital.

Previously, the patient was seen primarily in emergency care settings due to metabolic crises. Once connected with the pediatric mobile clinic program, not only was she able to receive primary care services, but she also gained access to community resources and assistance with navigating the complex medical system, with the hope that the patient will have regular visits with specialists instead of only being seen in acute crises. Delay in specialty and supportive care due to lack of health insurance, immigrant status, lack of financial resources and home instability lead to multiple hospitalizations and ultimately to the severe developmental delay noted in the patient. In the case of our patient, having had access to specialty care earlier on would've provided for better health outcomes for the patient. Delaying the treatment of severe LCHAD complications can lead to death.

Reference

LCHAD deficiency. Genetic and Rare Diseases Information Center . (2017, 12 17). LCHAD deficiency . Retrieved 1 16, 2018, from LCHAD deficiency : https://rarediseases.info.nih.gov/diseases/6867/lchad-deficiency