patients, UROS is defective only in a small number of myelodysplastic cells resulting from a somatic mutation in either *UROS* or *GATA1*. In that scenario, the number of clones carrying the pathogenic variant might be too low to be detected by standard molecular methods but sufficient to cause clinical and biochemical symptoms consistent with CEP.

Case presentation: The patient is a 52-year-old woman with a complicated past medical history who presented to a tertiary medical center with blistering lesions that started 2 years ago. Her past medical history is remarkable for type 2 diabetes, hypertension, hyperlipidemia, coronary artery disease, chronic obstructive pulmonary disease, and peripheral vascular disease.

She was diagnosed with CEP at her local clinic but had not had any medical interventions done locally. Over the years, her clinical condition deteriorated progressively where now she had multiple areas of ulceration and blistering with hypopigmented scarring in sun-exposed areas, particularly on her face and scalp, deep routed ulcers in her hands and arm, and reddish discoloration of her urine. In the preceding months, she had also developed worsening pancytopenia needing blood transfusion. A bone marrow biopsy showed erythroid hyperplasia with dyspoietic changes confined to the erythroid lineage and hypercellularity but no morphological evidence of malignancy. Molecular DNA analysis detected pathogenic variants in the genes *BCOR* and *TET2*, both associated with myelodysplastic syndrome and acute myeloid leukemia. Based on these findings, the patient was diagnosed with low-risk myelodysplastic syndrome.

Porphyrin analysis showed elevated levels of uro- and coproporphyrins in urine, type I porphyrin isomers in feces, and slightly increased total porphyrin in erythrocytes—a biochemical profile consistent with CEP. However, next-generation sequencing of 11 genes associated with porphyria did not detect any pathogenic variants, and the measured UROS activity in erythrocytes was within the normal range.

Conclusion: The case presented here is, to our knowledge, the first female patient reported with a late-onset CEP-like phenotype associated with myelodysplastic syndrome. This demonstrates the value of biochemical investigation and clinical examination in cases where DNA analysis proves inconclusive.

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Case presentation: Dual diagnosis of LCHAD deficiency and type 1 diabetes mellitus and complexities of management

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Background: Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency (OMIM # 609016) is an autosomal recessive of mitochondrial fatty acid β-oxidation caused by biallelic pathogenic variants in the *HADHA* gene. In this condition, fasting or catabolism, in the absence of adequate caloric intake, can result in hypoglycemia. Hypoglycemia leads to the utilization of free fatty acids, which then enter the mitochondria via the carnitine cycle. In individuals with a fully functional LCHAD enzyme, the hydroxy form of free fatty acids is oxidized to acetyl-CoA, which is then used to produce ketones; this process can compensate for the energy needs not satisfied by glucose. LCHAD Deficiency prevents adequate production of ketones and results in the accumulation of fatty acid intermediates that inhibit gluconeogenesis. The build-up of fatty acid intermediates is toxic. Features in untreated patients may include severe hypoketotic hypoglycemia, lethargy, liver dysfunction, hepatomegaly, metabolic acidosis, clotting abnormalities, hyperammonemia, cardiomyopathy, and sudden death. Treatment for LCHAD Deficiency includes avoidance of fasting, a diet limited in long chain fatty acids, and medium-chain triglyceride supplementation.

Type 1 Diabetes Mellitus (T1DM) is an autoimmune disorder that results in the destruction of pancreatic beta cells, which produce insulin. Insulin regulates the metabolism of carbohydrates, fats, and protein by promoting the absorption of glucose from the blood into liver, fat, and skeletal muscle cells. Treatment for T1DM includes exogenous insulin, diet, and exercise. Good glycemic control is required to reduce the risk of long-term sequelae of chronic hyperglycemia. One risk of strict glycemic control is the development of hypoglycemia.

The presence of both LCHAD Deficiency and T1DM in a single patient has not been reported to date.

Case presentation: We report our experience of management of a 10-year-old female with both LCHAD Deficiency and T1DM. She was diagnosed via newborn screening and managed at a different center until 6 years of age when she moved and transitioned care to our institution. Genetic testing revealed compound heterozygous variants in *HADHA* (NM_000182.4: c.[955 G>A];[c.499_501delACAinsCC]). She was treated in the standard fashion with avoidance of fasting, dietary management, and medium-chain triglyceride supplementation. She has struggled with compliance to her treatment regimen, particularly regarding the use of medium-chain triglyceride supplementation given the combination of taste, side effects and burden of supplementation.

At 9 years of age, she presented to her primary care physician's office with a several-week history of fatigue, abdominal pain, decreased appetite, and weight loss. Point of care blood glucose level was found to be 631. She was sent to the emergency department, where labs were remarkable for pH of 7.36 with a normal anion gap, HbA1c of 13.8, glucose and 20 ketones present in the urine, and significantly elevated glutamic acid decarboxylase antibody. She was admitted for stabilization and initiation of an insulin regimen for her new diagnosis of T1DM. Genetics was consulted in the hospital and recommended close monitoring of blood glucose levels, both in the hospital and upon discharge, given the risks associated with hypoglycemia in both conditions. Given the concern for hypoglycemia, a continuous glucose monitoring device was recommended, and continuous glucose monitoring was initiated after discharge from the hospital.

Her diet has been closely managed with assistance of both a metabolic dietician and diabetic dietician and requires close monitoring of both fat and carbohydrate intake. She and her family have struggled with compliance secondary to her many dietary restrictions and the complexity of management. She has not had any further hospitalizations since her diagnosis of T1DM.

Conclusion: Rare cases of T1DM in the setting of other fatty acid oxidation disorders (MCAD and VLCAD Deficiency) have been reported, but T1DM has not been reported in the setting of LCHAD Deficiency. To our knowledge, this is the first reported case of a patient diagnosed with both LCHAD Deficiency and T1DM. This case highlights the difficulty of managing these two coexisting conditions, as tight glycemic control in T1DM is necessary to prevent sequelae of hyperglycemia but comes with the risk of inducing hypoglycemia, which can result in a life-threatening metabolic crisis secondary to LCHAD Deficiency.

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