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Very Long Chain Acyl-CoA Dehydrogenase Deficiency Diagnosed by Michigan Newborn Screening and their Cardiac Complications

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Very Long Chain Acyl-CoA Dehydrogenase Deficiency Diagnosed by Michigan Newborn Screening and their Cardiac Complications

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With the expansion of newborn screening (NBS), there has been an increase in the number of patients diagnosed with genetic and metabolic conditions before the onset of symptoms. Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCADD) is a genetic disorder of fatty acid metabolism that can cause hypoglycemia, myopathy, rhabdomyolysis, cardiomyopathy and cardiac arrhythmias. VLCADD was included in the Michigan NBS in 2005 and since then, our center has diagnosed 28 patients with VLCADD. We have observed that many of these patients are asymptomatic or have a mild disease presentation. These patients may also have a lower incidence of cardiac complications than patients diagnosed from early-onset symptoms. VLCADD exhibits a wide range of clinical outcomes, depending on many factors including genotype, care access, and onset of symptoms. By reviewing current literature and our patient cohort data, we aimed to test our hypothesis that mild patients detected via NBS who are otherwise asymptomatic have a lower incidence of cardiac complications than previously suspected. Understanding this relationship may help optimize recommendations for treatment and screening in VLCADD patients.

Keywords: newborn screening, VLCADD, cardiac complications