Title: A rare case report on Clinico-genetic study of Medium Chain acyl-CoA Dehydrogenase (MCAD) deficiency in Pakistan

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Medium Chain Acyl-CoA Dehydrogenase (MCAD) deficiency is a metabolic disorder involving mitochondrial fatty acid oxidation that is inherited genetically. Essentially, this condition inhibits the body from breaking down specific lipids to produce energy. Metabolic issues may arise from an MCAD deficit. The defect in medium-chain acyl-CoA dehydrogenase was initially reported in 1982–1983. One of the enzymes involved in mitochondrial fatty acid β-oxidation is dehydrogenase. An enzyme called MCAD is involved in the medium-chain fatty acid oxidation process. It releases energy by catalyzing the first stage of these fatty acids' breakdown. A deficiency of MCAD can impact this first step of medium-chain fatty acid breakdown. As a result, partially oxidized fatty acid intermediates may accumulate in the body. Clinically, this may present as lethargy, metabolic acidosis, hypoketotic hypoglycemia, and potentially liver damage in affected individuals. If left untreated during periods of prolonged fasting when fatty acids are the primary fuel, the buildup of toxic intermediates can worsen symptoms and even lead to a life-threatening coma. This occurs as glycogen stores become depleted and the individual is unable to generate sufficient energy from alternate substrates. Early detection and management are therefore critical.

There is no previously reported case of MCAD in Pakistan. In this case report, three of the five girls in the family whose samples we received have now passed away. According to reports, two of them are still alive and exhibit no alterations at all. The third daughter, who lived for two months and her parents, provided the samples and data. Prior to her death from multiple organ failure, the patient underwent several tests and follow-ups. The third daughter's tests revealed a possible medium-chain acyl-CoA dehydrogenase impairment, which the doctors suspected after performing the core tests including blood CP, LFTs, RFTs and some serum tests. The clinical and genetic causes of MCAD have been determined using blood samples from the patient and the patient's family.