Joint Event

12th International Conference on
Mental Health and Human Resilience

9th International Conference on
Mental Health and Psychiatry

June 13-14, 2024

Rome, Italy

The first reported case of a child with two different rare metabolic disorders: Very Long-Chain acyl-CoA dehydrogenase deficiency and encephalomyopathic mitochondrial DNA depletion syndrome 13

Maha Alotaibi
King Saud Medical City, Saudi Arabia

One of the most common inborn errors in fatty acid β oxidation (FAO) is a very long-chain acyl-coenzyme A dehydrogenase (VLCAD) deficiency. It is autosomal recessive. The enzyme used in the first phase of FAO is VLCAD. The enzyme is responsible for βoxidation spiral pathway's initial step, the dehydrogenation process of long-chain fatty acyl-CoA. The phenotypes include hypoglycemia, hepatomegaly, cardiomyopathy, and occasionally abrupt mortality. Most VLCAD deficiencies in newborns are now detected during the neonatal period due to the development of newborn screening programs. Mitochondrial DNA depletion syndromes (MTDPS) are one of the rarest metabolic disorders. It is an autosomal recessive disease caused by defects in genes necessary for the maintenance of mitochondrial DNA (mtDNA). One of these FBXL4 (F-box and leucine-rich repeat protein 4) variants causes encephalomyopathic mtDNA depletionsyndrome 13 (MTDPS13), which presents as a failure to thrive, severe global develop-mental delay, hypotonia, early infantile onset of encephalopathy, and lactic acidosis. We report here the case of a Saudi infant born to consanguineous parents who presented to us with severe failure to thrive, profound neurodevelopmental delays, and facial dysmorphic features. Whole-exome sequencing (WES) showed the infants hadMTDPS13. The FBXL4 variant c.1698A > G p. (Ile566Met) has previously been described as a disease that causes developmental delay and lactic acidosis, and another variant has also been detected in the patient. The ACADVL variant c.134C > A p. (Ser45) has previously been described to cause VLCAD deficiency. A comprehensive literature review showed our patient to be the first case of MTDPS13 and VLCAD reported to date worldwide

Conclusion: This particular infant, the occurrence of these two uncommon metabolic disorders resulted in quite an unusual clinical presentation. The rise in "double trouble" instances shows that it is worthwhile to look for a more accurate diagnosis when an already established phenotype is compounded by unusual traits and difficult diagnosis.

Biography

Maha Alotaibi is a champion of genetic disease in ministry of health. She has plenty of researches in genetic and metabolic disorders and Ad-Hoc Reviewer. She is training and teaching the medical students from different medical collages and residents in Riyadh. She also participates as a speaker in international and local conferences