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Webinar

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Glutaric Aciduria Type II with Ketosis in a Male Infant

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Statement of the Problem: Glutaric aciduria type II (GA II) also known as multiple acyl-CoA dehydrogenase deficiency is an inborn metabolic disorder belonging to the family of organic acidurias. It is a disorder that interferes with the body's ability to break down proteins and fats to produce energy. The clinical presentation and onset of disease may vary depending upon the age, location, and nature of the mutations. Patients of GA-II present with varying symptoms like abnormal posturing, excessive crying and irritability, and clinical signs like hypotonia, tachycardia, tachypnoea, movement disorders, hypoglycaemia, and often neonatal death. Tandem mass spectrometry (TMS) acts as a screening tool, while the diagnosis of GA-II with ketosis is confirmed by a combination of tests like organic acids, quantitative random urine, and a full urine panel. Herein, we report a case of glutaric aciduria type-II with ketosis in a 4.5-month-old male infant who was managed with a low-protein diet, which was free of tryptophan, lysine, and other specific dietary supplements.

Conclusion & Significance: GA-II in the neonatal form with or without congenital anomalies is said to be associated with poor prognosis and high mortality. Early diagnosis, compliance to specialized diet, affordability, and regular follow-ups are required to tackle this potentially life-threatening condition.

Recent publications

1) Krutika Tandon, Rahul Tandon, Meet Patel, Charmy Parikh, and

Henil Upadhyay.

Glutaric Aciduria Type II with Ketosis in a Male Infant. *Cureus Journal* 2021. 1.7759/cureus.14407

2) Krutika Tandon, Rahul Tandon, Charmy G. Parikh, Henil M. Upadhyay, and Meet Patel.

Rare manifestation of common disease: An adolescent girl with transient ischemic attack (TIA) like presentation in multiple tuberculoma. *Journal of Family Medicine and Primary Care* 2021. 10.4103/jfmpc.jfmpc_603_21

Biography

Dr Meet Patel is an [NHS junior doctor](#) currently working as an SHO in Elderly Care, at Robert Jones & Agnes Hunt Orthopaedic Hospital (Oswestry, Shropshire). He has previously worked as an SHO in the Urology centre, at [Alexandra Hospital](#) (Redditch, Worcestershire). He has a background teaching work experience at Dr Mo Sobhy PLAB Academy (Liverpool, England) as a PLAB Tutor, Mentor & Mock Examiner and a Medicine tutor in Knowledge Institute of [Physiotherapy](#) (Gujarat, India). He is a global ambassador for Inspire Medics & Trewlink community (UK-based medical organizations). He completed his medical school and F1-level internship at Sardar Patel University (Gujarat, India).

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