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A family case report of tyrosinemia type-1 from Najran province of Saudi Arabia

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Tyrosinemia-1 is a very rare and severe inborn metabolic disorder affecting about 1 in 100,00 to 120,000 births worldwide. The pathophysiology of the disease is explained by the subsequent accumulation of tyrosine and its toxic metabolite in blood and tissues causing dysfunction of these organs, affecting mainly the liver and the kidney. The patient may develop acute liver failure in early infancy. The untreated survivors of the acute failure show liver cirrhosis, renal tubulopathy, rickets and hepatocellular carcinoma. The diagnosis of the disease depends on the clinical features and biochemical tests as detecting an increase in tyrosine and its metabolites in blood and urine. The main aim of this study was first; to report for the first time a family case of tyrosinemia in Najran city, the southern province of Saudi Arabia (KSA). Secondly; to increase the social and medical community awareness of the disease as consanguineous marriages are very common in this area.

Biography

Mohammed Chyad Hammoodi Al-Noaemi has completed his Ph.D. from University of Newcastle upon-Tyne (UK) in Physiology. Then after, he was involved in teaching physiology in more than 30 medical and paramedical colleges in Iraq, Sudan, Jordan, and Saudi Arabia. Currently, he is a professor of physiology in Al-Ghad International College in Najran-KSA. He has published about 50 papers in reputed journals.

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