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Early diagnosis in familial glucocorticoid deficiency

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Familial glucocorticoid deficiency (FGD) is a rare autosomal recessive condition, characterized by marked atrophy of zona fasciculata and reticularis with preservation of zona glomerulosa. Out of more than 50 published cases, 18 patients died as a result of glucocorticoid insufficiency. The main objective of this report is to emphasize the early diagnosis and treatment in our 17 month patient. Her presenting features following an upper respiratory tract infection were hypoglycemia, seizures as well as deep hyperpigmentation of the limbs and lips. A low cortisol concentration, elevated ACTH level and normal electrolytes and aldosterone level all supported the diagnosis of primary glucocorticoid deficiency. Parents were counseled about the diagnosis, management and the lifelong requirement of steroids. FGD is an easily treatable disease when recognized but frequently missed due to a non-specific presentation. FGD is a treatable disease, delayed diagnosis and treatment can lead significant morbid.

Biography

Fatima Hamad Al Jneibi has completed her MBBS from Faculty of Medical Health Sciences at UAE University in Al Ain, UAE and currently pursuing her Pediatric Residency at Sheikh Khalifa Medical City (SKMC), R4, Accreditation Council for Graduate Medical Education (ACGME) accredited program.

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