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Research Details :

Research Title	:	*Mitochondrial cytopathy presenting with features of Gitelman's syndrome* *Mitochondrial cytopathy presenting with features of Gitelman's syndrome*
Descriptipn	:	Mitochondria are essential for the homeostasis of every cell except red blood cells.1 Therefore, mitochondrial disorders cause a wide range of clinical presentations; however, organs with higher aerobic metabolism tend to be more severely affected.1 These symptoms, although severe, can be non-specific. Gitelman syndrome (GS) is a primary renal tubular disorder with hypokalemic metabolic alkalosis, hypocalciuria, and magnesium deficiency.2 Gitelman et al described it in 1966, in 3 female patients, 22- 47 years old. It is more typical of adults and age at presentation is usually 5 years or more. Failure to thrive and short stature has been described occasionally, and it could be included as an association.3 However, recently 3 cases of GS and growth hormone (GH) deficiency were reported,4,5 and considered as a new phenotype of GS with a new complex hereditary renal-tubular-pituitary syndrome.5 We report another case of GS that was associated with GH deficiency, partial adrenocortical hormone (ACTH) deficiency, and mitochondrial encephalopathy
Research Type	:	Article
Added Date	:	Monday, March 31, 2008

Researchers :

Researcher Name (Arabic)	Researcher Name (English)	Researcher Type	Degree	Email
أ.د/ محمد بن محمد سعيد جان		Researcher	أستاذ	

Attatchments :

File Name	Type	Description
Mitochondrial_Cytopathy.pdf	pdf	مشاهدة المقالة العلمية كاملة