HDR SYNDROME (BARAKAT SYNDROME): CASE REPORT

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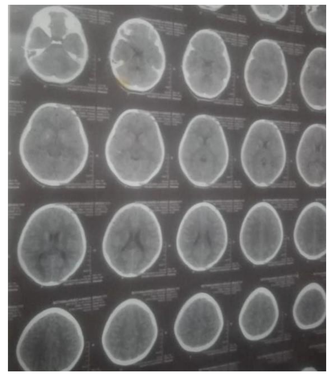
INTRODUCTION

- Barakat syndrome (HDR syndrome) components are hypoparathyroidism (H), sensorineural deafness (D) and renal disease (R)
- It caused by an autosomal dominant inheritance, being mostly associated with deletions in chromosome 10p14 or mutations in GATA3 gene.
- We present a girl with HDR syndrome in Alexandria university

CASE REPORT

- Here we describe an eleven years old girl
- she was born to non-consanguineous parents.
- She came to an emergency department complaining of the occurrence of one attack of tonic convulsion with loss of consciousness.
- Physical examination on admission revealed positive spasm of the feet and hands, she wears hearing aids.
- Laboratory assessment revealed
- Low total serum calcium (5.2 mg/dL, reference value (RV): 8.8 to 10.8mg/dL)
- Low parathyroid hormone (PTH) concentration (9.12pg/mL, RV: 9 to 52pg/mL)
- High serum phosphorus (10 mg/dL ,RV: 4-7 mg/dL)
- Magnesium (2.1 mg/dL, RV 1.7 to 2.7
- High Alkaline phosphatase (385 U/L, RV 46-116).

- The abdominal ultrasound reported a simple cyst with a thin wall and clear content in mid zone diffuse increase in cortical echogenicity of the kidneys.
- The audiogram revealed bilateral severesensorineural hearing impairment.
- CT brain revealed normal morphological features of both cerebral hemisphere and absent basal ganglia's calcificat
- Treatment was initiated with calcitriol and calcium carbonate supplementation.



CONCLUSIONS

- The combination of hypoparathyroidism congenital and sensorineural deafness and in pointed to the diagnosis of HDR syndrome

REFERENCES

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