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A CASE OF (HDR SYNDROME) BARAKAT SYNDROME WITH AN ASSOCIATED VITAMIN D DEFICIENCY

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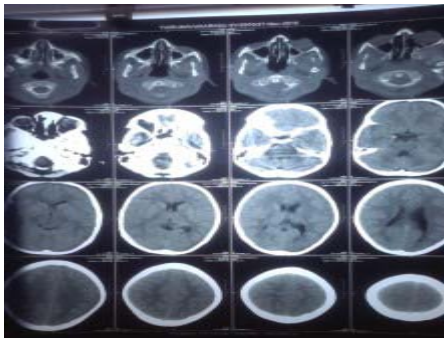
Abstract : HDR syndrome (Hypoparathyroidism, Deafness, Renal dysplasia) is a rare syndrome initially reported by Barakat in 1977. It is a clinical entity characterized by the triad of hypoparathyroidism, nerve deafness and renal dysplasia caused commonly by mutation of GATA 3 gene located at chromosome 10p15(1). Exact incidence is not known but it is a very rare condition with only few cases reported in literature (2). Here we report a 9years old boy who presented with recurrent afebrile seizures. Further investigations showed hypocalcaemia and hypoparathyroidism in the child. Imaging of the abdomen showed abnormalities of the kidney but renal parameters were normal. Audiometry showed mild sensorineural hearing loss thus confirming our diagnosis.

Keyword : Hypoparathyroidism,Sensorineural deafness, Renal dysplasia

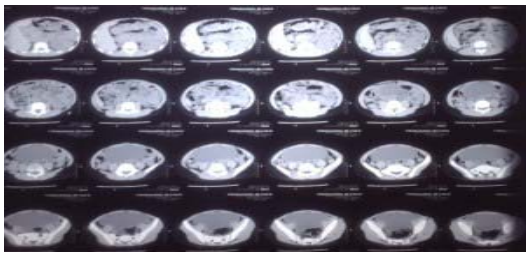
INTRODUCTION: Hypoparathyroidism, sensorineural deafness and renal disease was first described as syndrome called HDR syndromeby Amin Y. Brakat in 1977. He first described this syndrome in two male siblings who died at five years and other at three years. The clinical findings were confirmed by their autopsy findings[3]. Since then few more cases were reported in literature with varying associations. It usually follows autosomal dominant form of inheritance though similar clinical presentations following autosomal recessive patterns have been described. The defect in the majority of cases has been mapped to chromosome 10p (Gene Map Locus: 10pter-p13 or 10p14- p15.1) Hapaloinsufficiency (deletions) of zinc-finger transcription factor GATA3 or mutations in the GATA gene3 appear to be the underlying cause of this syndrome. It causes the failure in the specification of prosensory domain and subsequently leads to increased cell death in the cochlear duct thus causing deafness. Since the spectrum of phenotypic variation in affected patients is quite large, Barakat (HDR) syndrome probably arises as a low penetrance haploinsufficient disorder in which the patients' genetic background plays a major role in the severity of the disease. The GATA3 gene involved in this syndrome is a transcription factor involved in embryonic development of parathyroid glands, kindneys, inner ears, thymus and central nervous findings[4].

CASE REPORT: Nine years old male child came with complaints of one episode of generalized tonic clonic seizures. His past history suggested similar such episodes of seizures not associated with fever since age three. There was also history of recurrent episodes of carpopedal spasms, especially at the time of seizures. He was the first born of nonconsanguinous marriage with one younger male sibling of 7 years of age. No similar complaints in the family and his sibling was healthy. At the time of admission child was having active seizures with carpopedal spasms. Seizures were terminated with lorazepam and calcium correction was given for the child. His sensorium regained back to normal. Vital signs were normal. Neurological examination was also normal.



CT BRAIN

Rest of the systemic examination was normal. ECG and ECHO was taken which was normal. Laboratory investigations showedurea:18mg/dl,creatinine:0.7mg/dl, s.calcium:5mg, s.phosphorous:6, s.magnesium 1.5(normal range: 1.7-2.1mg/dl),s sodium: 143meq/l,s.potassium :3.2meq/l,vitamin D-8.40ng (deficiency 20ng/ml,insufficiency (20-29ng/ml),sufficiency (>30ng/ml).Intact parathyroid hormone -7.5pg/ml (ref value 15-68.3 pg/ml). There was no history of thyroid surgery or exposure to radiation. Arterial blood gas analysis was done which showed ionized calcium 0.69mmol/L(normal range 1.15-1.29).No other abnormalities were revealed in the ABG. Urine creatinine and urine magnesium was done which was normal. Urine culture was also normal. Neuroimaging was initially done for the seizures. CT brain which was done did not show any abnormalities.



CT ABDOMEN

USG abdomen showed non visualized Right kidney. Furthermore CT abdomen was also done which showed Reflux nephropathy ,contracted right kidney with compensatory hypertrophy of left kidney. No evidence of hydronephrosis in both kidneys were seen. Other organs were normal. MRI abdomen with urogram showed Right kidney severly contracted 4.2cm, Left kidney showed compensatory hypertrophy measuring 8.8*5cm. No evidence of hydrouretronephrosis. Bladder moderately distended ,shows diffuse increase in wall thickening (7mm) and shows multiple internal trabeculations. Micturating cystourethrogram was done which was normal. ENT evaluation was done. Pure Tone Audiometry showed mild sensorineural hearing loss in speech frequencies. He was discharged on vitamin D3 and calcium supplementation with high calcium diet. Also advised to avoid ototoxic drugs and exposure to high noises and yearly follow up in ENT. Nephrology follow up was also advised. Paediatric Surgery advised frequent voiding of bladder.

CONCLUSION: Diagnosis is based on the clinical findings. Diagnosis of suspected patients may be assisted by the following tests: measurement of PTH levels, an audiogram or auditory brain stem response study, renal imaging studies, and a renal biopsy. DNA analysis may demonstrate the presence of a submicroscopic deletion on chromosome 10p. Detailed study of chromosome 10p should be undertaken in patients with well-defined renal tract abnormality phenotypes, especially when there is associated hypoparathyroidism or deafness . Various combinations of this syndrome have been reported including familial idiopathic hypoparathyroidism and progressive sensorineural deafness without renal disease and autosomal recessive hypoparathyroidism with renal insufficiency and developmental delay. Treatment consists of treating the clinical abnormalities associated with hypoparathyroidism, deafness, and renal disease at the time of diagnosis[5]. Renal disease decides the main prognosis of the disease which includes nephrotic syndrome, renal dysplasia, hypoplasia or aplasia, pelvicalcalyceal deformity, vesicoureteral reflux, chronic kidney disease and renal scarring.

Hence child should be regularly followed up by a nephrologist since progression of renal disease is possible .His condition also is a case of HDR syndrome which has presented with an associated Vitamin D deficiency[6].Since the disease classically follows an autosomal dominant pattern his sibling should be screened though he is healthy and asymptomatic at present. Hence a high index of suspicion can pick up this very rare yet fascinating condition.

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