SYNDROME OF HYPOPARATHYROIDISM, DEAFNESS AND RENAL DISEASE (HDR) IN A 7 YEARS OLD BOY AT BAHAWALPUR

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ABSTRACT

Syndrome of (hypoparathyroidism, deafness and renal disease) HDS is a rare autosomal dominant syndrome which is characterized by triad of hypoparathyroidism, sensorineural hearing loss and renal disease. The syndrome is caused by mutation of GATA3 gene on chromosome 10*p*. We report a case of 7 years old boy who presented in Bahawalpur Victoria Hospital Bahawalpur, Pakistan with recurrent tetanic spells, muscular aches and pains for last 15 days. He was operated four years back for renal stones. His blood complete picture was normal except for mild anemia. His serum calcium level was low i.e. 6.9 mg/dL. Serum magnesium, phosphate, alkaline phospha-tase and Vitamin D levels were normal. Serum PTH (parathyroid hormone) level was low i.e. 4.51 pg/ml (Normal range 11-67 pg/mL). Urine RE showed albumin, pus cells and calcium oxalate crystals. Pure tone audiometry showed bilateral mild to moderate degree of sensorineural hearing loss.

Keywords: Hypoparathyroidism, Renal disease, Sensorineural deafness.

INTRODUCTION

Hypoparathyroidism, deafness and renal disease (HDR) syndrome is also known as Barakat syndrome. It is a very rare autosomal dominant syndrome[1,2]. It was first described in 1977 by Barakat *et al*, reason for being called as Barakat syndrome as well[3]. The prevalence of this syndrome is still unknown with only around a dozen cases reported so far.The term HDR was coined for this syndrome by Bilous *et al*, who reported cases of this syndrome in a family in 1992[4]. We report the first case of HDR/Barakat syndrome in Pakistan who presented in Bahawal Victoria hospital Bahawalpur, Pakistan in March 2017.

CASE REPORT

The patient is 7 years old boy who presented in Bahawal Victoria Hospital Bahawalpur, Pakistan with recurrent tetanic spells, muscular aches and pains for last 15 days. His past history is suggestive of bilateral hearing impairment and was operated four years back for renal calculi. His developmental history was insignificant. On examination he had visible dysmorphism, malaligned teeth, short stature, left elbow contracture and mild scoliosis. His height was 109cm and weight was 17kgs both below the 50th centile. Rest of the systemic examination was normal. His BCP (blood complete picture) was normal except for mild anemia. His serum calcium level was low i.e 6.9 mg/dL. Serum PTH (parathyroid hormone) level was low i.e. 4.51 pg/ml (Normal range 11-67 pg/mL). Urine routine examination showed albumin, pus cells and calcium oxalate crystals. His ultrasound abdomen shows bilateral increased echogenicity in both kidneys but his renal diethylenetriamine pentacetate (DTPA) scan showed adequate functioning kidneys. There was no visible morphological malformation on Computerized Tomogram (CT scan) of abdomen except for butterfly vertebra at L1 (first lumber vertebra) leading to scoliotic deformity. His CT scan brain showed no calcifications. His electrocardiogram (ECG) and Echocardiography were normal. Pure tone audiometry (PTA) showed bilateral mild to moderate degree of sensorineural hearing loss. This triad of hypoparathyroidism, renal disease and nerve deafness constitutes HDR/Barakat syndrome. Serum calcium levels of parents and 3 siblings (sisters) were normal.

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DISCUSSION

HDR syndrome is a rare autosomal dominant syndrome with unknown prevalence. It is established that inheritance is autosomal dominant[1-4].

This syndrome is caused by mutation in GATA-3 gene which is located at 10*p*[5]. This particular region is involved in the development

Table: Laboratory investigations.

Test	Measured Level	Normal Value
Hb	10.7 g/dL	12-18 g/dL
TLC	7.92 X 10^3	4-11 X 10^3
Platelets	289 X 10^3	15-400 X 10^3
S. Calcium	6.9 mg/dL	8.8-10.8 mg/dL
S. Magnesium	2.2 md/dL	1.6-2.4 mg/dL
S. PO4 (phosphate)	6.8 mg/dL	3.2-5.8 mg/dL
S.PTH	4.51 pg/ml	11-67 pg/ml
BSR	91 mg/dL	<140 mg/dL
S.ALT	17 iu/L	<40 iu/L
S. Alkaline phosphatase	388 iu/L	65-306 iu/L
S.Urea	21 mg/dL	10-50 mg/dL
S.Creatinine	0.7 mg/dL	0.6-1.1 mg/dL
S. Sodium	138 mEq/L	135-155 mEq/L
S. Potassium	4.2 mEq/L	3.5-5.5 mEq/L
Blood pH	7.44	7.35-7.45
Urine RE		
pH	6	
Albumin	Trace	
Pus Cells	2-4	
Calcium Oxalate Crystals	1-2	

of embryonic parathyroid glands, kidneys and inner ear. That is why these patients have hypoparathyroidism, renal disease and hearing loss[5].

These patients can report to physicians with tetany, convulsion, hearing impairment, renal disease or any combination of these. 90% of patients with HDR syndrome have hypoparathyroidism and hearing loss, and 80% have renal dysplasia[1]. Hearing loss associated with HDR syndromeis sensorineural usually bilateral. Renal diseasecan vary from proteinuria, hematuria, nephrotic syndrome, cystic kidney, renal dysplasia, hypoplasia, aplasia, vesicoureteral reflux, pelvicalyceal deformity and scarring[3,4]. Hypoparathyroidism has been reported to appear earliest among the three symptoms (hypoparathyroidism, nerve deafness and renal disease) by Chenouard A *et al*[6].

The original cases described by Barakat *et al*, had presented with hypocalcaemia and proteinuria which gradually progressed to steroid resistant nephrotic syndrome[3]. They also had hypoparathyroidism and bilateral sensorineural hearing loss. All those four siblings died between 3 to 8 years of life. In 1992, Bilous *et al* reported HDR syndrome in two brothers and two daughters of one of the brothers[4]. In literature, very few cases of HDR syndrome have been reported so far[7]. First case was reported in 1977 by Barakat until recently Higuchi *et al* reported a case of HDR syndrome in association with biliary atresia in 2016[8].

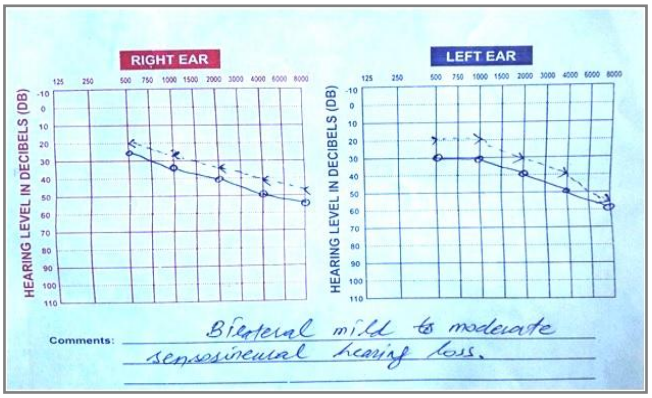

Figure: Audiogram of Child.

CONCLUSION

HDR syndrome should be suspected in any case presenting with hearing impairment, low serum calcium and renal disease.

CONFLICT OF INTEREST

This study has no conflict of interest to be declared by any author.

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