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# Barakat syndrome: Case report of a rare syndrome

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**Abstract**
Barakat syndrome, also known as HDR syndrome is an autosomal dominant disorder characterized by hypoparathyroidism, sensorineural deafness, and renal dysplasia caused by mutation of the GATA3 gene.The syndrome was first noted in siblings with hypocalcemia and proteinuria. It is a genetic developmental disorder with clinical diversity; affected people usually present with hearing loss, tetany, seizures, numbness and renal disease at any age. Hearing loss is usually bilateral and may range from mild to severe. Renal disease mainly includes renal-dysplasia, nephrotic syndrome, cystic kidney disease. Other reported features are intellectual disability, polycystic ovaries, distinct facial characteristics, ischemic stroke and retinitis pigmentosa. Treatment is symptomatic and prognosis depends upon the severity of renal disease.

**Keywords:** HDR syndrome, Barakat syndrome, Hypoparathyroidism, renal dysplasia

## Introduction
Barakat Syndrome is characterised by hypoparathyroidism, sensorineural deafness, and renal disease. However, specific symptoms and severity can vary. Around 65% of people with Barakat syndrome have hypoparathyroidism, sensorineural deafness and kidney disease together. HDR syndrome is caused by mutation of the GATA3 gene. GATA3 gene is expressed in parathyroid glands, inner ear, kidney, thymus, and central nervous system [1-5]. Ferraris et al. reported that 62.3% of patients diagnosed with HDR syndrome had classical triad of hypoparathyroidism, sensorineural deafness and renal disease while 28.6% had hypoparathyroidism and sensorineural deafness and only 2.6% had sensorineural deafness and renal disease together, and 6.5% patients had isolated deafness [6]. Both male and females have been noted to be affected. The renal disorders seen in this syndrome can present in various forms such as renal agenesis, renal dysplasia, hypoplasia, and vesicoureteral reflux [7, 8] and many patients progress to chronic renal failure [9]. Here we report a case of Barakat syndrome presented to us with seizure, hypoparathyroidism and bilateral sensorineural deafness.

## Case Report
A 21 years old male patient presented to us with bilateral hearing loss since birth and seizures which started at the age of 15 years and later on developed numbness in both hands. Physical examination was normal with average built and BMI of 21 kg/m$^2$. No facial dysmorphism was noted. Laboratory findings included low total serum calcium, high phosphorus with low serum intact parathyroid hormone (iPTH) levels (table 1). Non-contrast CT brain was normal without any signs of calcification of brain parenchyma. Abdominal ultrasonography and CT scan showed the absence of renal tissue on the left side s/o left renal aplasia. Pure tone audiometry revealed bilateral sensorineural hearing loss at high frequencies (figure 1).

**Table 1:** Laboratory Tests values

| Name of investigation | Observed value | Normal range | Interpretation |
|---|---|---|---|
| Serum Total Calcium | 6.2 mg/dl | 8.5-10.5 mg/dl | Low |
| Serum Phosphorus | 5.6 mg/dl | 2.5-4.5 mg/dl | High |
| S.Alkaline Phosphatase | 125 U/L | 50-580 U/L | Normal |
| S.Parathyroid Hormone | 6.5 pg/ml | 10-65 pg/ml | Low |
| Serum Vitamin-D | 42 ng/ml | 30-50 ng/ml | Normal |
| Serum Creatinine | 0.6 mg/dl | 0.5-1.5 mg/dl | Normal |

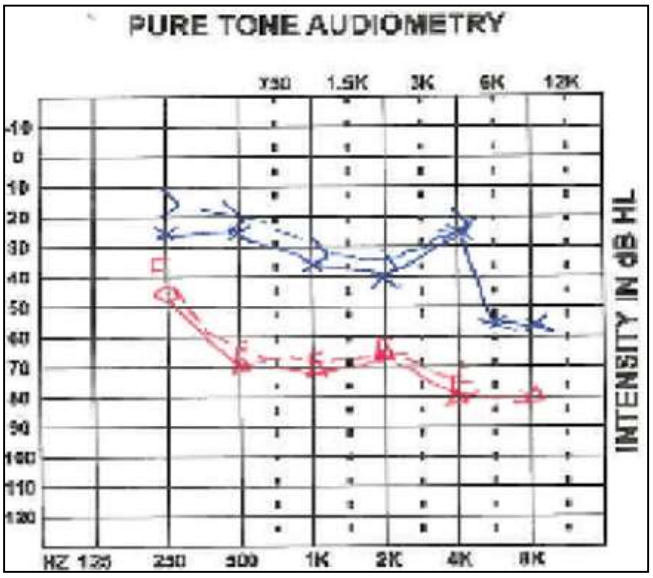


**Fig 1:** Pure tone audiometry findings showing sensorineural hearing loss

## Discussion

Based on the clinical and laboratory findings, the patient was diagnosed to have Barakat/HDR syndrome, which is characterized by hypoparathyroidism, sensorineural deafness, and renal agenesis. HDR syndrome can present at any age. Our case had hypocalcemic symptoms like seizure and numbness with bilateral hearing loss at the time of presentation. But unfortunately, genetic analysis could not be performed in our case due to financial restraints. Calcifications, caused by hypoparathyroidism, commonly occur primarily in basal ganglia, but may also occur in the thalamus, dentate nuclei, cerebral cortex, and centrum semiovale [5, 7]. Hypoparathyroidism can present as asymptomatic hypocalcemia or hypocalcemic tetany, paresthesia (numbness), and seizures. Serum levels of parathyroid hormone (PTH) can range from low normal to undetectable levels. Hypoparathyroidism often cause neurological and psychiatric disorders including vertigo, impaired cognitive function, and depression[5]. Hearing loss in HDR syndrome is usually more severe at the higher end of the frequency spectrum [7]. Our patient had mild and bilateral sensorineural hearing loss at high frequencies with left renal agenesis. Treatment of patients with this syndrome should be comprehensive and should include genetic counseling. Management is essentially symptomatic and depends on the clinical findings and severity of the disease. Hypocalcemia is usually the most common problem requiring treatment. Deafness should be diagnosed and treated early with hearing amplification, and if needed cochlear implantation. The treatment of kidney disease depends on the abnormality present. Prognosis depends upon the nature and severity of kidney disease. Patients with minor kidney disease are expected to have a normal life expectancy. Based on the findings of the present case it is concluded that patients presenting with seizures associated with deafness and renal disorder should be evaluated carefully and Barakat syndrome should be kept in mind while doing the work up.

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