

Hyperkalemia Unveiled: A Case of Barakat Syndrome

Here we present an interesting case of Barakat syndrome with resistant hyperkalemia due to hyporeninemic hypoaldosteronism (Type 4 renal tubular acidosis). Barakat or HDR syndrome is an autosomal dominant disorder that consists of triad of hypoparathyroidism, sensorineural deafness, and renal dysplasia.^[1,2] As per our knowledge, we report the first case of Barakat syndrome presenting with resistant hyperkalemia and convulsions.

A 39-year-old female was admitted with history of recurrent episodes of generalized seizures due to hypocalcemia despite being on oral calcium and sodium valproate and easy fatigability for 3 months. Neuroimaging was normal. Further history revealed progressive hearing loss for last 6 years with bilateral moderate sensorineural hearing loss on audiometry.

Laboratory parameters revealed blood urea of 60 mg/dl, serum creatinine 1.7 mg/dl, serum potassium 6.6 mEq/L, chloride 115 mEq/L serum calcium 3.9 mg/dl, albumin 4.6 g/dl, serum bicarbonate 18 mEq/L, serum anion gap 12 mEq/L (normal 8-16 mEq/L) and phosphorus 10.5 mg/dL. Electrocardiography demonstrated prolonged QT interval (0.52 sec) and tall T waves. Ultrasound abdomen showed a solitary right kidney with raised echogenicity. Further investigations yielded very low PTH of 0.23 pg/ml (normal range 15-65 pg/ml) by electrochemiluminescence immunoassay (ECLIA) and high TSH 22 mIU/l (normal range 0.4-4 mIU/l). Her urine examination revealed 770 mg of proteinuria in 24 hours urine collection. There was no history of diabetes, hypertension, fever, neck surgery or irradiation. There was no significant family history.

Initially, she was managed with anti-hyperkalemic treatment in the form of dextrose insulin drip, furosemide, oral bicarbonate supplements, polystyrene exchange resins and low potassium diet but continued to have hyperkalemia (potassium 6.1 mEq/L). A possibility of hyporeninemic hypoaldosteronism was considered based on normal anion gap hyperchloremic metabolic acidosis and low transtubular potassium gradient of 2 (normal >4). However, her serum aldosterone level was normal. She was started on fludrocortisone 100 µg daily and within 2 days her potassium decreased to 4 mEq/L. Renal involvement, hypoparathyroidism and sensorineural deafness completed our clinical triad of Barakat syndrome. Genetic mutation analysis could not be done due to financial constraints. The patient was continued on maintenance dose of fludrocortisone (50 µg/day), calcium carbonate, thyroxine, ergocalciferol, calcitriol and sodium valproate. She is maintaining normal levels of potassium and calcium with no further seizure episode.

Barakat syndrome (HDR syndrome) is caused by haploinsufficiency of the GATA3 gene on chromosome 10p15 and patients with this syndrome usually have initial symptoms related to hypocalcemia.^[3] It was first reported in 1977 by Barakat *et al.*^[1] GATA3 belongs to GATA transcription factor families, which are involved in vertebrate embryonic development of the parathyroid glands, inner ears, kidney, thymus, and central nervous system. Hypoparathyroidism causes symptomatic or asymptomatic hypocalcemia by normal or inappropriately low levels of parathyroid hormone (PTH) secretion. Sensorineural deafness is a consistent feature, which is more severe at higher frequencies and usually bilateral.^[1,2] Severity of renal involvement also varies from no renal abnormalities to renal dysplasia, hypoplasia, cystic kidneys, vesicoureteral reflux and most patients progress to advanced chronic kidney disease requiring renal replacement therapy.^[4] Diagnosis is generally based on clinical findings, though DNA analysis will aid in definitive diagnosis by showing haploinsufficiency or submicroscopic deletion on chromosome 10p.^[1,3] Treatment of this syndrome is symptomatic, treating the clinical abnormalities associated with HDR syndrome. Prognosis of HDR syndrome is varied depending on the nature and severity of the renal disease.^[4]

This is an unusual presentation with hyperkalemia. This has not been reported in any previous case reports, though renal tubular acidosis (type 1 and 2) have been reported earlier too.^[5] There are case reports involving multiple endocrine organ involvement.^[5] This patient too was having associated thyroid involvement. Hyperkalemia was responding to empirical fludrocortisone so probably it was due to type 4 Renal Tubular acidosis (hyporeninemic hypoaldosteronism) along with early renal dysfunction.

To conclude, Barakat or HDR syndrome should be suspected in a patient with deafness, hypocalcemia and renal dysfunction and one should always think of hyporeninemic hypoaldosteronism in situation of unexplained hyperkalemia with mild renal dysfunction.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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