Barakat Syndrome, also known as HDR Syndrome, is a rare genetic disorder characterized by the triad of hypoparathyroidism (H), neurosensory deafness (D) and renal disease (R) and caused by mutations in GATA3 on chromosome 10p14-p15.

**CASE REPORT**

ELJ, male, 9 years old, was admitted at emergency service complaining of pain and contractures on upper and lower limbs which initiated four months earlier and got worse in the last week. Furthermore, he presented significant weight loss in the period.

**Birth data:**
Gestational age 42 weeks, weight 3080g, lenght 46cm, cephalic perimeter 33cm, apgar 9.

**Physical Exame:**
Regular general condition, mild dehydration, bilateral palpebral edema
Weight: 20kg (P<3), Height 116cm (P<3), PH: 164.0 cm (<P3)
Capillary blood glucose = 96 mg/dL
Hypertonia and muscle contracture of upper and lower limbs. Painful mobilization of hips and of hands and feet small joints.
Negative Trousseau and Chvostek’s sings
Pubertal stage: Tanner I

**Exams:**
Admissional (table 1); after iniciate treatment (table 2).
Abdominal ultrasound: kidneys with diffuse increase in cortical echogenicity.
Audimetry: sensorineural deafness

**Treatment:** Teriparatide, calcium carbonate, calcitriol, hydrochlorothiazide, Scholl’s solution and elemental phosphorus.

**DISCUSSION/ CONCLUSION**

The prevalence of HDR syndrome is unknown and presents a variable phenotypic expression (HDR, HD, DR, HR, R and D). Neurosensory deafness is the most common component of the syndrome (96.7%), hypoparathyroidism (93.3%) and kidney disease (72.2%). Diagnosis is based on clinical symptomatology and molecular analysis.

In the present case, the patient show all three components of the syndrome. Renal disease presented as severe tubulopathy. Because hypocalcemia was unresponsive to calcium plus calcitriol replacement, Teriparatide (rh-PTH 1-34) was initiated twice a day. After 3 years of follow up, patient progressed with hearing impairment, reduced bone mineral density (fig.1), incipient nephrocalcinosis without deterioration of renal function and severe short stature (fig. 2).

Diagnosis and management of HDR syndrome is still challenging.

**REFERENCES**