BARAKAT SYNDROME (HDR SYNDROME): CASE REPORT

MARINA BRESSIANI, ANGÉLICA DALL'AGNESE*, ADRIANA GODINHO, CÉSAR GEREMIA, MARCIA PUÑALES*
Pediatric Endocrinology Service, Institute for Children with Diabetes, Conceição Children Hospital,
Conceição Hospital Group - Ministry of Health,
Porto Alegre, Rio Grande do Sul, Brazil

INTRODUCTION

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 GATA 3 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.
Audiometry: sensorineural deafness

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

Table 1

Exame	Result	Reference Value
PTHi	2.38 pg/mL	15–65 pg/mL
Calcium	5.59 mg/dL	8,2–10.3 mg/dL
Phosphorus	1.4 mg/dL	4–7mg/dL
Creatinine	1.17 mg/dL	0.4–0.6 mg/dL
Glycosuria	693mg/dL	não detectável
Calciuria	0.72 mg/mg	<21 mg/mg
Albuminuria	41.8 mg/L	<29 mg/L
Glomerular Filtration Rate	68ml/min/1.73 m^2	>90ml/min/1,73 m^2

Table 2

EXAMS (2018)	March	July	Reference Values
CO2 (mEq/L)	17	19	21-32
Blood Glucose (mg/dL)	85	88	70-99
Creatinine (mg/dL)	0.9	0.88	0.4-1.2
Urea (mg/dL)	34	25	10-50
Calcium (mg/dL)	8.8	9.1	8.2-10.3
Magnesium (mg/dL)	1.8	1.8	1.7-2.5
Phosphorus (mg/dL)	4.9	5.3	4.0-7.0
Potassium (mEq/L)	4.0	4.0	3.5-5.0
Sodium (mEq/L)	140	138	135-145
25-hidroxyvitamin D	-	19.1	30-60
Glycosuria	367	268	-
Urinary magnesium (mg/dL)	8	8	-
Urinary potassium (mEq/L)	48	28	-
Urinary sodium (mEq/L)	41	65	-
Albuminuria (mg/L)	39	-	< 29
Calciuria (mg/dL)	17	19.6	-
Uricosuria (mg/dL)	28	29	-
Creatininuria (mg/dL)	54	51	-

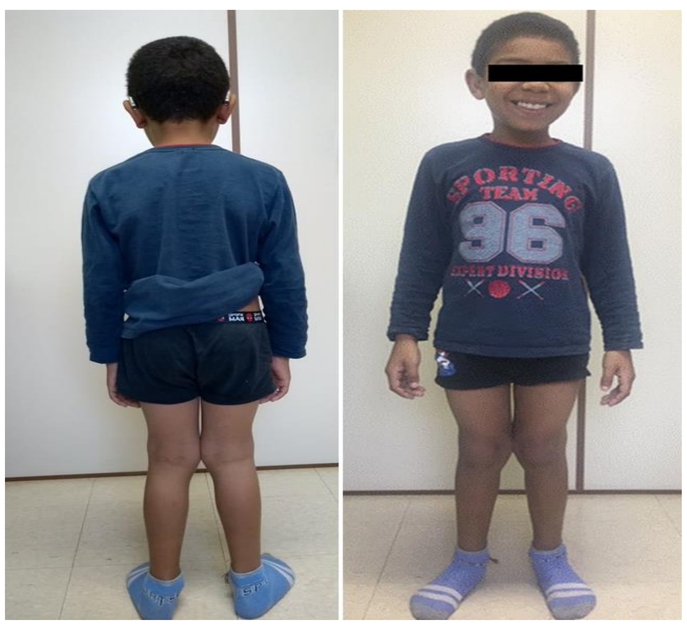


fig.1

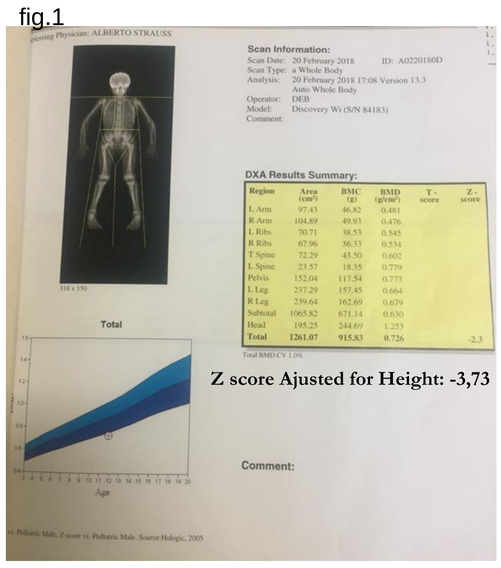
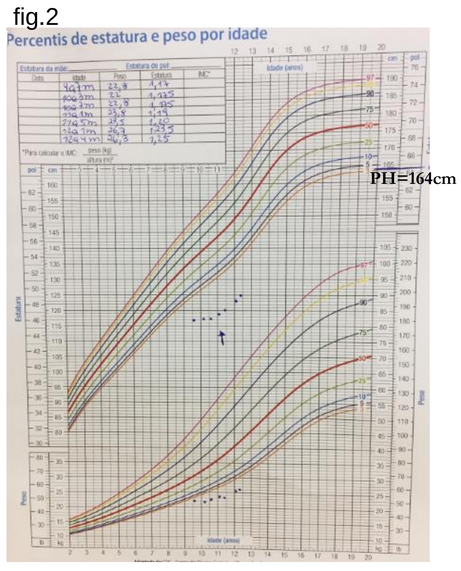


fig.2



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 managment of HDR syndrome is still challenging.

REFERENCES

1.Carpenter, T. Etiology of hypocalcemia in infants and children. Up to date. 2017, Aug 10th
2.Yang, A. et. al. HDR syndrome with a novel mutation in *GATA3* mimicking a congenital X-linked stapes gusher: a case report. BMC Med Genet. 2017; 18: 121.
3.Gogorza MS, Mena E, Serra G, Jiménez A, Noval M, Pereg V. Síndrome hypoparathyroidism-deafnessrenal dysplasia: descripción de un caso. Endocrinol Diabetes Nutr. 2018;65:187---188.
4.Mutlu, G. A Novel De Novo GATA Binding Protein 3 Mutation in a Turkish Boy with Hypoparathyroidism, Deafness, and Renal Dysplasia Syndrome. J Clin Res Pediatr Endocrinol. 2015 Dec; 7(4): 344–348.