Conflicts of interest

The authors declare that they have no potential conflicts of interest related to the contents of this article.

REFERENCES

1. Mulliez SM, de Keyser F, Verbist C, Vantilborgh A, Wijns W, Beukinga I, et al. Lupus anticoagulant-hypoprothrombinemia syndrome: report of two cases and review of the literature. Lupus. 2015;24:736–45.
2. Rapaport SI, Ames SB, Duvall BJ. A plasma coagulation defect in systemic lupus erythematosus arising from hypoprothrombinemia combined with antiprothrombinase activity. Blood. 1960;15:212–27.
3. Bajaj SP, Rapaport SI, Fierer DS, Herbst KD, Schwartz DB. A mechanism for the hypoprothrombinemia of the acquired hypoprothrombinemia-lupus anticoagulant syndrome. Blood. 1983;61:684–92.
4. Raflores MB, Kaplan RB, Spero JA. Pre-operative management of a patient with hypoprothrombinemia-lupus anticoagulant syndrome. Thromb Haemost. 2007;98:248–50.
5. Mazodier K, Arnaud L, Mathian A, Costedoat-Chalumeau N, Haroche J, Frances C, et al. Lupus anticoagulant-hypoprothrombinemia syndrome: report of 8 cases and review of the literature. Medicine. 2012;91:251–60.

Gonzalo Carreño-Tarragona[a], Enrique Morales[b,*], María Carmen Jiménez-Herrero[b], Elena Cortés-Fornieles[b], Eduardo Gutierrez[b], Manuel Praga[b]

[a] *Servicio de Hematología, Hospital Universitario 12 de Octubre, Madrid, Spain*
[b] *Servicio de Nefrología, Hospital Universitario 12 de Octubre, Madrid, Spain*

[*] *Corresponding author.*
E-mail address: emoralesr@senefro.org (E. Morales).

http://dx.doi.org/10.1016/j.nefroe.2015.11.008

Barakat syndrome or HDR syndrome: Another association of kidney disease and deafness☆

Síndrome hipoparathyroidism, deafness and renal displasia o síndrome de Barakat otra asociación de sordera y nefropatía

Dear Editor,

If a patient has kidney disease and deafness, we think on Alport syndrome, a widespread entity. However, this is not always the case. One of the recent issues of the journal Nefrología included an excellent review of kidney disease in the context of mitochondrial diseases and how nephrologists should suspect this diseases in a nephropathy (tubulopathy or glomerular injury, manifested by kidney failure and proteinuria) which is accompanied by hearing loss or sensorineural deafness.[1] In this letter, we present a case of Barakat syndrome or hypoparathyroidism, deafness and renal dysplasia (HDR) syndrome, another disease that should be included in the differential diagnosis of kidney disease and hereditary deafness.[2,3]

A 32-year-old patient was admitted to the obstetrics department owing to oedema. She was 36 weeks pregnant and the laboratory tests revealed a plasma creatinine at 1.4 mg/dl and 6 grams of proteinuria in a 24-h urine collection. Medical history included familial hypoparathyroidism in chronic treatment with vitamin D and calcium carbonate, bilateral sensorineural deafness and left kidney agenesis. With this history, in 2007, she was diagnosed with HDR syndrome. A genetic study demonstrated the presence of the c.431 mutation in the GATA3 gene (gene for the transcription factor GATA3, located in the short arm of chromosome 10). Both the patient and her mother were heterozygous for this mutation. The rest of the family (father, sister and maternal aunt) did not have any clinical manifestations of the syndrome; nevertheless, a molecular genetic study ruled out the presence of this mutation. Prior to pregnancy, the patient had been examined in the urology department as she was a single-kidney patient. She had undergone laboratory testing that showed mild kidney failure with a serum Cr of 1.3 mg/dl and proteinuria at 2.8 g/day.

Since she did not have HTN, the presence of pre-eclampsia was ruled out, and she was diagnosed with worsening of renal function in a patient with chronic kidney disease (CKD) in pregnancy.

☆ Please cite this article as: Rodríguez Benítez P, Jaldo Rodríguez MT, Hernández Coronado A, Torres Aguilera E, Melero R, Tejedor A. Síndrome hipoparathyroidism, deafness and renal displasia o síndrome de Barakat otra asociación de sordera y nefropatía. Nefrologia. 2016;36:188–189.

Given the severe and persistent proteinuria with the progressive decline in kidney function, it was decided to perform a Caesarean section, which proceeded without complications. After birth, she was started on antiproteinuric treatment with enalapril (10 mg/day). Currently (2 years after the birth), the patient has stage-3 CKD, with plasma creatinine at 1.7 mg/dl, glomerular filtration rate estimated by the MDRD-4 equation at 40 ml/min and albuminuria at 74 mg/day.

HDR syndrome or Barakat syndrome is a rare autosomal dominant hereditary disease determined by a mutation in the GATA3 gene, a gene located in the short arm of chromosome 10.[4] This gene is a transcription factor involved in the embryonic development of the parathyroid glands, kidneys, inner ears, thymus and central nervous system. This syndrome is characterised by the classic triad of familial primary hypoparathyroidism, with the possible occurrence of seizures, as a manifestation of hypocalcaemia, renal dysplasia and various degrees of sensorineural deafness. Most cases are bilateral.[5]

From a nephrology point of view, it may manifest with hypoplasia, renal dysplasia or agenesis, pyelocalyceal deformity, renal cysts, vesicoureteral reflux, renal tubular acidosis, chronic kidney failure (sometimes requiring renal replacement therapy), haematuria and various degrees of proteinuria. Nephrotic syndrome, which is typically resistant to steroids, may occur. The most common finding on kidney biopsy is focal segmental glomerulosclerosis.[6]

Treatment is symptomatic and aimed at managing or treating the clinical abnormalities associated with hypoparathyroidism, deafness and kidney disease, essentially, management of proteinuria.

Barakat syndrome is an uncommon cause of proteinuric nephropathy that should be suspected in cases associated with hypoparathyroidism and hereditary sensorineural deafness. Early diagnosis and treatment with renin–angiotensin system blockers may help slow the progression of kidney failure and improve prognosis, which essentially depends on the severity of kidney failure.

Conflicts of interest

The authors have no conflicts of interest to declare.

REFERENCES

1. Cavero T, Rabasco C, Molero A, Blázquez A, Hernández E, Martín MA, et al. ¿Cuándo debe sospechar un nefrólogo una enfermedad mitocondrial? Nefrologia. 2015;35:6–17.
2. Barakat AY, D'Albora JB, Martin MM, Jose PA. Familial nephrosis, nerve deafness and hypoparathyroidism. J Pediatr. 1977;91:61–4.
3. Bilous RW, Murty G, Parkinson DB, Thakker RV, Coulthard MG, Burn J, et al. Autosomal dominant familial hypoparathyroidism, sensorineural deafness and renal displasia. New Eng J Med. 1992;327:1069–74.
4. Van Esch H, Groenen P, Nesbit MA, Schuffenhauer S, Lichtner P, Vanderlinden G, et al. GATA3 haplo-insufficiency causes human HDR syndrome. Nature. 2000;406:419–22.
5. Maleki N, Bashardoust B, Alamdari MI, Tavosi Z. Seizure, deafness and renal failure: a case of Barakat syndrome. Case Rep Nephrol. 2013;2013:261907.
6. Taslipinar A, Kebapcilar L, Kutlu M, Sahin M, Aydogdu A, Uckaya G, et al. HDR syndrome (hypoparathyroidism, sensorineural deafness and renal disease) accompanied by renal tubular acidosis and endocrine abnormalities. Intern Med. 2008;47:1003–7.

Patrocinio Rodríguez Benítez[a,*], María Teresa Jaldo Rodríguez[b], Andrés Hernández Coronado[a], Esther Torres Aguilera[a], Rosa Melero[a], Alberto Tejedor[a]

[a] *Servicio de Nefrología, Hospital General Universitario Gregorio Marañón, Madrid, Spain*
[b] *Servicio de Nefrología, Complejo Hospitalario de Jaén, Jaén, Spain*

[*] *Corresponding author at*: Servicio de Nefrología, Hospital General Universitario Gregorio Marañón, C/ Dr. Esquerdo, 46, 28007 Madrid, Spain. Tel.: +34 915868319/915868674.
E-mail address: prodriguezb@senefro.org (P. Rodríguez Benítez).

http://dx.doi.org/10.1016/j.nefroe.2015.04.006