HDR syndrome in a Colombian woman with a genital tract malformation: First case report in Latin America

Síndrome HDR en una mujer colombiana con una malformación genitourinaria: Primer reporte de caso en Latinoamérica

Michael A. Vallejo-Urrego, Alejandra M. Parra-Morales y Adriana González

Received 15th March 2018 / Sent for Modification 19th July 2018 / Accepted 11th August 2018

ABSTRACT

Objectives Hypoparathyroidism, sensorineural deafness and renal disease (HDR) syndrome, also known as Barakat syndrome, is an autosomal dominant transmission hereditary disease with a wide range of penetrance and expressivity. Haploinsufficiency of the GATA3 two finger zinc transcription factor is believed to be its cause. This is the first time this orphan disease is reported in Latin America, so the publishing of this report is expected to raise awareness on these types of syndrome, that are usually underdiagnosed in our region, which in turn causes an increase in the years lost to disability (YLDs) rates, as well as higher costs to be assumed by public health systems.
Methods A 36-year-old Colombian woman diagnosed with parathyroid gland agenesis was referred from the Endocrinology Service to the Outpatient Service. According to her medical record, in the past she had developed hypocalcaemia, left renal agenesis, hypoparathyroidism, bicornate uterus and sensorineural hearing loss. Through a genetic analysis a pathological mutation on the short arm of the GATA 3 gen (c.404dupC, p Ala136 GlyfsTER 167) was confirmed, which led to a HDR syndrome diagnosis.
Discussion This case proves that there is a possibility that mutations described in other continents may be developed by individuals from our region. Regardless of ethnicity, Barakat syndrome should be considered as a possible diagnosis in patients presenting the typical triad that has been described for this condition, since there could be underdiagnosis of this disease in Latin-America due to the lack of knowledge on this condition in said region, and that genetic counseling in these patients is of great importance for the implications of the syndrome in future generations.

Key Words: Hypoparathyroidism; deafness; gata3 transcription factor; uterine cervical disease; zinc fingers *(source: MeSH, NML).*

RESUMEN

Objetivos El síndrome de hipoparatiroidismo, sordera neurosensorial y displasia renal (HDR) también llamado síndrome de Barakat, es una enfermedad hereditaria de transmisión autosómica dominante con amplia penetrancia y expresividad genética. El síndrome es causado por la haploinsuficiencia del factor de transcripción de dedos de Zinc GATA3. Esta es la primera vez que esta enfermedad huérfana es reportada en latinoamerica, y buscamos generar consciencia de la presencia de estas enfermedades, las cuales usualmente son infradiagnósticadas en nuestro medio y llevan a un aumento de años perdidos por discapacidad y costos para el sistema de salud pública.
Métodos Una mujer colombiana de 36 años ingresó a consulta externa de genética referida por el servicio de endocrinología por una agenesia de paratiroides. La paciente tenía antecedentes de hipocalcemia, agenesia renal izquierda, hipoparatiroidismo, sordera neurosensorial y útero bicorneo. Se realizó un análisis genético que confirmo una mutación patológica en el brazo corto del gen GATA3 (c.404dupC, p Ala136 GlyfsTER 167) diagnóstica del síndrome de Barakat.

MV: MD. M. Sc. Human Genetics, Specialist in Epidemiology, Specialist in Quality Management; Genetics Department, Sociedad de Cirugía de Bogotá, Hospital de San José, Fundación Universitaria de Ciencias de la Salud (FUCS). Bogotá, Colombia.
mvallejo@hospitaldesanjose.org.co
AP: MD. Mandatory Social Service in progress in Research activities; Research Division, Sociedad de Cirugía de Bogotá, Hospital de San José, Fundación Universitaria de Ciencias de la Salud (FUCS). Bogotá, Colombia.
alejandramariamd@gmail.com
AG: MD. Internal medicine Specialist, Endocrinology Fellow; Endocrinology Division, Sociedad de Cirugía de Bogotá, Hospital de San José, Fundación Universitaria de Ciencias de la Salud (FUCS). Bogotá, Colombia.
adripatgonzalez@hotmail.com

DOI: https://doi.org/10.15446/rsap.V20n5.71057