Genetics and Molecular Research

eNOS gene Glu298Asp and 4b/a polymorphisms are associated with renal function parameters in Mexican patients with Fabry disease

A. Marin-Medina[1,2], A.J.L. Brambila-Tapia[3], V.J. Picos-Cárdenas[4], M.P. Gallegos-Arreola[2] and L.E. Figuera[1,2]

[1]Doctorado en Genética Humana, Instituto de Genética Humana, Centro Universitario de Ciencias de la Salud, Universidad de Guadalajara, Guadalajara, Jalisco, México
[2]División de Genética, Centro de Investigación Biomédica de Occidente, Instituto Mexicano del Seguro Social, Guadalajara, Jalisco, México
[3]Departamento de Fisiología, Centro Universitario de Ciencias de la Salud, Universidad de Guadalajara, Guadalajara, Jalisco, México
[4]Laboratorio de Genética Humana, Unidad de Investigación, Facultad de Medicina, Universidad Autónoma de Sinaloa, Sinaloa, México

Corresponding author: L.E. Figuera
E-mail: luisfiguera@yahoo.com

Genet. Mol. Res. 15 (4): gmr15047802
Received December 17, 2015
Accepted January 15, 2016
Published October 24, 2016
DOI http://dx.doi.org/10.4238/gmr15047802

ABSTRACT. Fabry disease (FD) is an inherited X-linked lysosomal disease that causes renal failure in a high percentage of affected individuals. The *eNOS* gene encodes for endothelial nitric oxide synthase, which plays an important role in glomerular hemodynamics. This gene has two main polymorphisms (Glu298Asp and 4b/a) that have been studied in the context of many different diseases, including those involving cardiovascular and renal alterations. Considering the

lack of information regarding *eNOS* variants and FD, we investigated whether there were associations between *eNOS* genetic variants and renal function parameters in Mexican patients with FD and renal impairment. In total, 15 FD patients with renal alterations were included in the present study, and associations between *eNOS* polymorphisms and renal function parameters (urea, creatinine, and GFR) were evaluated. The Asp298 and 4a alleles of the *eNOS* gene were found to be significantly associated with increased levels of urea and creatinine, and a decreased glomerular filtration rate in FD patients, and this association behaved in a co-dominant fashion. Our results coincide with previous reports showing an association between these polymorphisms and kidney disease, and along with other studies regarding their role in the nitric oxide pathway, suggest that these variants affect the severity of nephropathy in patients with FD.

Key words: *eNOS* polymorphisms; Glu298Asp; 4b/a (VNTR); Fabry disease; Renal function parameters; Kidney disease