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Endomyocardial biopsy finding in an end-stage renal disease patient with c.196G>C in the α-galactosidase A gene

Yura Chae, Jongho Son, Eun Sil Koh, Sungjin Chung
Department of Internal Medicine-Nephrology, The Catholic University of Korea, Yeouido St. Mary's Hospital, Korea, Republic of

Case Study: Background: Among many α-galactosidase A (GLA) gene mutations causing Fabry disease, c.196G>C (p.E66Q) variant has been thought to be a functional polymorphism because residual enzymatic activity is relatively preserved and a few pathological studies of renal, cardiac and skin tissues from subjects with c.196G>C have found no accumulation of globotriaosylceramide (Gb3). Contrary to previous findings, this report provides evidence that the c.196G>C mutation in the GLA gene could be a pathologic polymorphism.
Case presentation: A 42-year-old female visited our clinic for recurrent hypotension and chest discomfort. She had been on peritoneal dialysis since 2013 for end-stage renal disease secondary to presumed diabetic kidney disease. In 2018, umbilical hernia required the transfer from peritoneal dialysis to permanent hemodialysis. Cardiac workup revealed that she had positive tilt table testing diagnostic for vasovagal syncope. Although the patient had no family history of Fabry disease, mild left ventricular hypertrophy on transthoracic echocardiography led to performing enzymatic and genetic analysis of GLA. The residual GLA activity was slightly low (32.3 μmol/h/mg protein) and a missense mutation c.196G>C was detected on exon 2 in the GLA gene. Despite normal finding on cardiac MRI, electron microscopic examination of endomyocardial biopsy specimen revealed a few round whorled laminated structures filing myocyte cytoplasm, consistent with Fabry disease. The patient had no other clinical manifestations such as angiokeratoma of skin, acroparesthesia and cornea verticillate.
Conclusion: These findings show that some patients with c.196G>C mutation may have pathological findings of Fabry disease and an endomyocardial biopsy for diagnostic purposes could be considered especially in patients with genetic variants of unknown significance and left ventricular hypertrophy.