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Name of journal: World Journal of Clinical Cases

ESPS Manuscript NO: 14690

Columns: Case Report

Advanced Anderson-Fabry disease presenting with left ventricular apical aneurysm and ventricular tachycardia

Marie-France Poulin, Alap Shah, Richard G Trohman, Christopher Madias

Abstract

A 54 year-old female with Anderson-Fabry disease (AFD) - R342Q missense mutation on exon 7 in alpha-galactosidase A (GLA) gene - presented with sustained ventricular tachycardia. Imaging confirmed the presence of a new left ventricular apical aneurysm (LVA) and a significantly reduced intra-cavitary gradient compared to two years prior. Anderson-Fabry disease is an X-linked lysosomal storage disorder caused by GLA



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