

## ESPS PEER-REVIEW REPORT

**Name of journal:** World Journal of Clinical Cases

**ESPS manuscript NO:** 14690

**Title:** Advanced Anderson-Fabry Disease Presenting with Left Ventricular Apical Aneurysm and Ventricular Tachycardia

**Reviewer's code:** 00227348

**Reviewer's country:** Japan

**Science editor:** Xiu-Xia Song

**Date sent for review:** 2014-10-20 19:47

**Date reviewed:** 2014-10-23 17:58

CLASSIFICATION	LANGUAGE EVALUATION	SCIENTIFIC MISCONDUCT	CONCLUSION
<input type="checkbox"/> Grade A: Excellent	<input checked="" type="checkbox"/> Grade A: Priority publishing	PubMed Search:	<input type="checkbox"/> Accept
<input checked="" type="checkbox"/> Grade B: Very good	<input type="checkbox"/> Grade B: Minor language polishing	<input type="checkbox"/> The same title	<input checked="" type="checkbox"/> High priority for publication
<input type="checkbox"/> Grade C: Good	<input type="checkbox"/> Grade C: A great deal of language polishing	<input type="checkbox"/> Duplicate publication	<input type="checkbox"/> Rejection
<input type="checkbox"/> Grade D: Fair	<input type="checkbox"/> Grade D: Rejected	<input checked="" type="checkbox"/> Plagiarism	<input type="checkbox"/> Minor revision
<input type="checkbox"/> Grade E: Poor		<input checked="" type="checkbox"/> No	<input type="checkbox"/> Major revision
		BPG Search:	
		<input type="checkbox"/> The same title	
		<input type="checkbox"/> Duplicate publication	
		<input type="checkbox"/> Plagiarism	
		<input checked="" type="checkbox"/> No	

## COMMENTS TO AUTHORS

Authors showed a case of AFD complicated by apical aneurysm and ventricular tachycardia. This report is written and presents well. Q1 12-lead ECG after termination of VT was not shown. Apical aneurysm developed at least during last 2 years. Serial ECGs should be presented. In HCM, ST-elevation in leads V4-6 often appears during the course of developing aneurysm. Q2 Apical aneurysm is associated with thrombus formation. In addition, the patient had atrial fibrillation. Was anticoagulation therapy performed? Q3 Were antiarrhythmic drugs prescribed such as amiodarone or sotalol?

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**Name of journal:** World Journal of Clinical Cases

**ESPS manuscript NO:** 14690

**Title:** Advanced Anderson-Fabry Disease Presenting with Left Ventricular Apical Aneurysm and Ventricular Tachycardia

**Reviewer's code:** 01293596

**Reviewer's country:** Japan

**Science editor:** Xiu-Xia Song

**Date sent for review:** 2014-10-20 19:47

**Date reviewed:** 2014-10-28 17:42

CLASSIFICATION	LANGUAGE EVALUATION	SCIENTIFIC MISCONDUCT	CONCLUSION
<input type="checkbox"/> Grade A: Excellent	<input type="checkbox"/> Grade A: Priority publishing	PubMed Search:	<input type="checkbox"/> Accept
<input type="checkbox"/> Grade B: Very good	<input type="checkbox"/> Grade B: Minor language polishing	<input type="checkbox"/> The same title	<input type="checkbox"/> High priority for publication
<input type="checkbox"/> Grade C: Good	<input type="checkbox"/> Grade C: A great deal of language polishing	<input type="checkbox"/> Duplicate publication	<input type="checkbox"/> Rejection
<input type="checkbox"/> Grade D: Fair	<input type="checkbox"/> Grade D: Rejected	<input type="checkbox"/> Plagiarism	<input type="checkbox"/> Minor revision
<input type="checkbox"/> Grade E: Poor		<input type="checkbox"/> No	<input type="checkbox"/> Major revision
		BPG Search:	
		<input type="checkbox"/> The same title	
		<input type="checkbox"/> Duplicate publication	
		<input type="checkbox"/> Plagiarism	
		<input type="checkbox"/> No	

## COMMENTS TO AUTHORS

General comments:

1. The formation of left ventricular apical aneurysm is quite rare in Fabry disease. Additionally, in female patients, the onset of symptoms is usually later compared with male patients. However, this patient presented with skin lesions, hypohydrosis and limb pain in her childhood. I wonder those findings including mid-ventricular obstruction are the characteristics of the VR342Q missense mutation in exon.
2. The authors suggested a shared mechanism of LVAA formation in Fabry disease and HCM. Are there any previous reports suggesting a shared mechanism for LVH or apical aneurysm?

Specific comments:

1. The 12-lead ECG after cardioversion should be exhibited as a figure.
2. The patient manifested various symptoms which are typical for Fabry disease. I think enzyme replacement therapy should have been started earlier.



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3. How is the involvement of kidney or central nervous system? I concern that apical aneurysm was caused by embolization due to atrial fibrillation. Was she treated with anticoagulation drugs?
4. The first paragraph in page 5 should be cited at 'Case presentation'.

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**Name of journal:** World Journal of Clinical Cases

**ESPS manuscript NO:** 14690

**Title:** Advanced Anderson-Fabry Disease Presenting with Left Ventricular Apical Aneurysm and Ventricular Tachycardia

**Reviewer's code:** 02887986

**Reviewer's country:** Turkey

**Science editor:** Xiu-Xia Song

**Date sent for review:** 2014-10-20 19:47

**Date reviewed:** 2014-10-21 16:04

CLASSIFICATION	LANGUAGE EVALUATION	SCIENTIFIC MISCONDUCT	CONCLUSION
<input checked="" type="checkbox"/> Grade A: Excellent	<input checked="" type="checkbox"/> Grade A: Priority publishing	PubMed Search:	<input checked="" type="checkbox"/> Accept
<input type="checkbox"/> Grade B: Very good	<input type="checkbox"/> Grade B: Minor language polishing	<input type="checkbox"/> The same title	<input type="checkbox"/> High priority for publication
<input type="checkbox"/> Grade C: Good	<input type="checkbox"/> Grade C: A great deal of language polishing	<input type="checkbox"/> Duplicate publication	<input type="checkbox"/> Rejection
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<input type="checkbox"/> Grade E: Poor		<input checked="" type="checkbox"/> No	<input type="checkbox"/> Major revision
		BPG Search:	
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		<input type="checkbox"/> Duplicate publication	
		<input type="checkbox"/> Plagiarism	
		<input checked="" type="checkbox"/> No	

## COMMENTS TO AUTHORS

Comments to the Author 1. TITLE: The title is suitable and covers the correct data of the presented case. 2. ABSTRACT: The abstract is completely sufficient. 3. FIGURES: The data are very well presented in the figures. 4. PATIENTS, MATERIAL AND METHODS: The methodology part of the paper is clear and makes the case presentation understandable. 5. RESULTS AND DISCUSSION The presentation and interpretation of the results is perfect. I congratulate the authors' team for diagnosis and presentation of the case.

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**Name of journal:** World Journal of Clinical Cases

**ESPS manuscript NO:** 14690

**Title:** Advanced Anderson-Fabry Disease Presenting with Left Ventricular Apical Aneurysm and Ventricular Tachycardia

**Reviewer's code:** 01576314

**Reviewer's country:** Brazil

**Science editor:** Xiu-Xia Song

**Date sent for review:** 2014-10-20 19:47

**Date reviewed:** 2014-11-04 20:40

CLASSIFICATION	LANGUAGE EVALUATION	SCIENTIFIC MISCONDUCT	CONCLUSION
<input type="checkbox"/> Grade A: Excellent	<input type="checkbox"/> Grade A: Priority publishing	PubMed Search:	<input type="checkbox"/> Accept
<input type="checkbox"/> Grade B: Very good	<input checked="" type="checkbox"/> Grade B: Minor language polishing	<input type="checkbox"/> The same title	<input type="checkbox"/> High priority for publication
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		<input type="checkbox"/> Duplicate publication	
		<input type="checkbox"/> Plagiarism	
		<input checked="" type="checkbox"/> No	

## COMMENTS TO AUTHORS

The manuscript by Marie-France Poulin et al, "Advanced Anderson-Fabry Disease Presenting with Left Ventricular Apical Aneurysm and Ventricular Tachycardia" is a Fabry disease case report where the authors describe the presence of a missense mutation (R342Q) in a 54 years old woman with strong cardiac phenotype. The manuscript is concise, well written and brings an important aspect of Fabry mutations in women. Despite the authors are not the first to describe apical hypertrophy in Fabry patients (Francisca Caetano et al, 2014 - Fabry disease presenting as apical left ventricular hypertrophy in a patient carrying the missense mutation R118C), they are the first to show such a strong heart alteration. Before the manuscript is accepted, some review is necessary. Criticisms - the authors should describe what is known about this mutation (R342Q) in the literature regarding pathogenicity. This mutation is present in the HGMD databank and some authors have already described it before and even checked its impact in functionality assays (Wu et al, Human Mutation 2011). Please cite and discuss these findings. - The authors should better describe Fabry disease in women. This is an important issue today due to the fact that women present later symptoms when



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compared to men and the difficulty to diagnose them. - It is not clear when the patient received the recombinant enzyme for the first time. Has the patient received regularly the treatment during this period? Which dose she received and how often? Was she followed during this time? How did the disease progress during this time? What was the ejection fraction of this patient two years ago? - Is the patient hypertensive? Has she any other cardiovascular complication? - The authors should better contextualize hypertrophic cardiomyopathy. - The text lack on important references regarding important issues like the ones cited before.