



SDI Review Form 1.6

Journal Name:	<u>British Journal of Medicine and Medical Research</u>
Manuscript Number:	Ms_BJMMR_32156
Title of the Manuscript:	Screening for Fabry Disease among Dialysis Patients in Brazil: Findings from the First 18 months of a Nationwide Study
Type of the Article	Original Research Article

General guideline for Peer Review process:

This journal's peer review policy states that **NO** manuscript should be rejected only on the basis of '**lack of Novelty**', provided the manuscript is scientifically robust and technically sound.

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PART 1: Review Comments

	Reviewer's comment	Author's comment <i>(if agreed with reviewer, correct the manuscript and highlight that part in the manuscript. It is mandatory that authors should write his/her feedback here)</i>
<u>Compulsory</u> REVISION comments	<p>The raw results reported in the article are interesting: the enzymatic and/or genetic analysis was performed in almost 3000 patients, and there are only few studies reported in literature with this high number of studied subjects. However, in the paper there are some mistakes that need to be corrected:</p> <ol style="list-style-type: none"> 1) The author designed an algorithm that allowed them to identify Fabry patients. Even if they wrote that this algorithm needs to be validated (page 18, last line), the enzymatic and/or genetic analysis was not performed in patients that were excluded by the algorithm. Therefore, it is possible that Fabry patients could be present also in the group of "not-selected subjects". For this reason, according to my opinion the results should be reported as "descriptive" study, without the intent to create an algorithm for the identification of Fabry subjects (until validation). Moreover, the percentage of Fabry patients identified using this algorithm is uncorrect (see point 2), and this could influence its validity. 2) According to your interpretation of the results, you considered patients with GLA mutations as affected by Fabry disease. It is true that Fabry disease is caused by mutations in this gene, but NOT ALL the mutations are responsible for the disease. <i>E.g.</i> R118C is considered as functional polymorphism (Ferreira S et al, 2015); the pathological meaning of S126G in Fabry disease is still unclear, and so on. For this reason, these mistakes need to be corrected before publication. 	
<u>Minor</u> REVISION comments	The manuscript will need to be reviewed for grammatical typographical errors.	



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	Moreover, some figures are too small and it is difficult or impossible to read what it is written (e.g.: fig.3).	
<u>Optional/General</u> comments	The raw results presented in this paper are excellent. However, the article presented some mistakes (the intent of the algorithm, the analysis of the GLA mutations), and I strongly recommend the publication of the paper after their correction.	

Reviewer Details:

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