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SDI FINAL EVALUATION FORM 1.1

PART 1:

Journal Name:	**British Journal of Medicine and Medical Research**
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 Article:	**Original Research Article**

PART 2:

FINAL EVALUATOR'S comments on revised paper (if any)	**Authors' response to final evaluator's comments**
In their paper, the authors considered some mutations in the GLA gene as responsible for FD, even if their role in the disease is still unknown/controversial. The authors supported the causative role of mutation citing some databases without consulting the most important ones for FD like Fabry-database.org, and the databases that they used are not-well "consulted" (e.g. : S126G : they cited, among the databases, the NCBI VarClin. According this database, the pathological meaning of S126G is not clear). However, if they want to demonstrate that a patient in whom they found a controversial mutation is affected , they should perform the Lyso-Gb3 analysis, which was not carried out. The definition of mutation as pathological or not is not a trivial issue, because it means that a patients will be treated or not with ERT, and this is an ethical issue for doubt mutations . **Moreover, since there is no validation, the algorith proposed by the authors is not scientifically robust.** **In conclusion, the previous requests are still not satisfied**	

Reviewer Details:

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