



PEER-REVIEW REPORT

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Title: Novel α -galactosidase A gene mutation in a Chinese Fabry disease family: A case report

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Reviewer's code: 03366151

Position: Peer Reviewer

Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: Italy

Author's Country/Territory: China

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Reviewer chosen by: AI Technique

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Scientific quality	<input type="checkbox"/> Grade A: Excellent <input type="checkbox"/> Grade B: Very good <input checked="" type="checkbox"/> Grade C: Good <input type="checkbox"/> Grade D: Fair <input type="checkbox"/> Grade E: Do not publish
Language quality	<input type="checkbox"/> Grade A: Priority publishing <input checked="" type="checkbox"/> Grade B: Minor language polishing <input type="checkbox"/> Grade C: A great deal of language polishing <input type="checkbox"/> Grade D: Rejection
Conclusion	<input type="checkbox"/> Accept (High priority) <input type="checkbox"/> Accept (General priority) <input checked="" type="checkbox"/> Minor revision <input type="checkbox"/> Major revision <input type="checkbox"/> Rejection
Re-review	<input checked="" type="checkbox"/> Yes <input type="checkbox"/> No



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Peer-reviewer statements	Peer-Review: [<input checked="" type="checkbox"/>] Anonymous [<input type="checkbox"/>] Onymous Conflicts-of-Interest: [<input type="checkbox"/>] Yes [<input checked="" type="checkbox"/>] No
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SPECIFIC COMMENTS TO AUTHORS

In this manuscript Fu et al. report on a novel α -galactosidase A gene mutation (348delG:p.G116fs) in exon 2 in a Chinese Fabry disease family. This is an interesting, clearly written paper. Minor comments The authors should underscore the delay for diagnosis and point out that a lysosomal storage disease should have been suspected years ago in the presence of cardiac hypertrophy, arrhythmias, and renal involvement. Family history (two close relatives with same heart disorder) should have reinforced the suspicion of a genetic nature of disease. Table 1 shows that the proband and her 52 years old sister had a normal (<10 mm) thickness of left ventricular (LV) posterior wall in spite of an increased thickness of interventricular septum (16 mm and 15.8 mm, respectively). Thinning of LV posterior wall is a feature of Fabry related cardiomyopathy in the late stage. The authors could address this point in discussion. Please use a galactosidase- α and a galactosidase - β instead of aglubberase- α and aglubberase- β , on page 11. As concerning proband's son, there is discrepancy between the value of LV posterior wall reported in the text and that shown in table 1. Please check and amend.