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LETTER TO THE EDITOR

Response to: No strong evidence to date for an association between RIMS1 and retinal dystrophy: Mahrood O, et al.

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Dear Editor,

We thank Drs Mahrood et al. for bringing to our attention their latest findings in relation to the seminal report implicating pathogenicity of *RIMS1*. Their conclusion that *RIMS1* lacks an evidentiary base as a monogenic cause of retinal dystrophy, is reassuring for our patient, who in most respects displays a profile of incomplete congenital stationary night blindness [1]. We note however the presence of unusually depressed scotopic responses in our case. Whilst this may reflect heterogeneity within the *CACNA1F*-related disease spectrum [2], the possibility of allelic modifiers is also acknowledged. Raising the question as to whether the identified variance in *RIMS1* is phenotypically complicit or simply an innocent bystander; and emphasising the importance of interrogation of potential modifying factors across the inherited retinal dystrophies.

References

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2. Men CJ, Bujakowska KM, Comander J, Place E, Bedoukian EC, Zhu X, Leroy BP, Fulton AB, Pierce EA (2017) The importance of genetic testing as demonstrated by two cases of *CACNA1F*-associated retinal generation misdiagnosed as LCA. Mol Vis 10(23):695–706

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