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REPLY TO LETTER

Reply to comment on: A novel dysferlin mutant pseudoexon bypassed with antisense oligonucleotides

Janice A. Dominov[1], Özgün Uyan[1], Peter C. Sapp[1], Diane McKenna-Yasek[1], Babi Ramesh Reddy Nallamilli[2], Madhuri Hegde[2] & Robert H. Brown Jr[1]

[1]Neurology Department, University of Massachusetts Medical School, Worcester, Massachusetts
[2]Department of Human Genetics, Emory University School of Medicine, Atlanta, Georgia

Correspondence Tel: 508-334-1271; Fax: 508-856-4485; E-mail: Robert.Brown@umassmed.edu

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We were delighted to read the comments from our colleague, Dr. Martin Krahn and his team,[1] noting that they, too, detect the deep intronic dysferlin mutation that we described.[2] We fully agree that this will likely be an infrequent mutation, even in the population of dysferlinopathy cases with only one known exonic dysferlin mutation, though it might be recurrent in specific populations. We anticipate that there will be other mutations deep within introns and other regulatory regions that will be identified through expanded strategies to screen for mutations such as the RNA analyses described in our paper, which could be applied to screens for other diseases as well.

Conflict of Interest

J. A. D. and R. H. B. Jr. are employed by the University of Massachusetts Medical School and co-inventors on a pending patent application: "COMPOSITIONS AND METHODS FOR MODULATING DYSFERLIN EXPRESSION". Dr. Brown reports grants from Cecil B. Day Foundation, grants from NIH-NINDS, and other from Howard Hughes Medical Institute.

References

1. Kergourlay V, Blandin G, Blanck V, et al. Comment on: a novel dysferlin mutant pseudoexon bypassed with antisense oligonucleotides. Ann Clin Transl Neurol 2015.
2. Dominov JA, et al. A novel dysferlin mutant pseudoexon bypassed with antisense oligonucleotides. Ann Clin Transl Neurol 2014;1:703–720.