Our work reporting one cause of MPSI, inherited mutations of SLC25A22, certainly falls within this context. The observation of Dr Striano and colleagues of 21 SLC25A22-negative cases from among their patients with MPSI is consistent with the many cases we also reported in which we did not find mutations in this gene.

Given this established genetic heterogeneity, the approach they suggest for the evaluation of an individual patient with MPSI is logical, with evaluation first for KCNT1 and then for other associated genes. We would include in such an evaluation MPSI is logical, with evaluation first for KCNT1 and then for other associated genes. We would include in such an evaluation

POTENTIAL CONFLICTS OF INTEREST

Nothing to report.

REFERENCES


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Questions about Efficacy of Exon-Skipping Therapy for Duchenne Muscular Dystrophy

Satyakam Bhagavati, MD

Mendell et al1 report strikingly positive results after treatment of Duchenne muscular dystrophy (DMD) boys with eteplirsen, an exon-skipping antisense morpholinol oligonucleotide. The number of dystrophin-positive fibers in muscle biopsy specimens was reported to increase (compared to pretreatment levels) by up to 52% after 48 weeks. Treatment for 48 weeks also showed a statistically significant improvement in walking ability