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Targeted Treatments for Inherited Neuromuscular Diseases of Childhood.

Fay AJ1, Knox R2, Neil EE3, Strober J1.

Author information

1 Department of Neurology, University of California San Francisco, San Francisco, California.
2 Department of Neurology, Ohio State University, Columbus, Ohio.
3 Department of Pediatrics, University of Michigan, Ann Arbor, Michigan.

Abstract

In the past decade, the number of genes linked to neuromuscular diseases of childhood has expanded dramatically, and this genetic information is forming the basis for gene-specific and even mutation-specific therapies. At the forefront of these advances are the two recently approved treatments for spinal muscular atrophy: one, an antisense oligonucleotide that modifies splicing of the SMN2 gene, and, the other, a gene therapy vector that delivers the SMN1 gene to motor neurons, both of which are allowing patients to acquire developmental milestones previously unseen in this fatal disease. This review highlights these advances and emerging targeted therapies for Duchenne muscular dystrophy and centronuclear myopathy, while also covering enzyme replacement therapy and small molecule-based targeted therapies for conditions such as Pompe's disease and congenital myasthenic syndromes. With these and other newer techniques for targeted correction of genetic defects, such as CRISPR/Cas9, there is now hope that treatments for many more genetic diseases of the nervous system will follow in the near future.

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Conflict of interest statement

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