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Gene therapy for paediatric muscular dystrophies: Revolutionizing treatment and future directions

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Background: Muscular dystrophies (MDs) are progressive neuromuscular disorders causing severe disability and premature mortality in children. Traditional treatments have focused on symptom management, but gene therapy offers a paradigm shift. Techniques like adeno-associated viral (AAV) vector-based treatments and CRISPR-Cas9 gene editing aim to correct the dystrophin gene defect, potentially slowing disease progression. This review synthesizes recent advancements in gene therapy for paediatric MDs, evaluates current clinical trials, and explores future directions.

Methods: A systematic review of peer-reviewed studies, clinical trial data, and meta-analyses published in the last decade was conducted. Databases searched include PubMed, Cochrane Library, Ovid, Elsevier, ClinicalTrials.gov, and Google Scholar. Studies on AAV-mediated microdystrophin gene therapy, exon-skipping, and CRISPR-Cas9 were included.

Results: Clinical trials have shown promising results for gene therapy in MDs. AAV-mediated microdystrophin therapy (e.g., SRP-9001) demonstrated significant improvements in dystrophin expression and motor function. Exon-skipping approaches restored functional dystrophin in patients, improving ambulation and muscle strength.

Discussion: CRISPR-Cas9 gene editing is emerging as a promising tool for genetic correction, with studies showing dystrophin restoration through exon skipping or gene deletion correction. Challenges include immune responses to viral vectors, off-target effects, and long-term safety.

Conclusion: Gene therapy represents a revolutionary approach for paediatric MDs, potentially slowing disease progression and improving quality of life. Early-phase trials are promising, though further research is needed to optimize delivery, minimize immunogenicity, and refine gene editing. Ethical concerns regarding genome modification in children should also be addressed.

Biography

Patrick Reardon is a final-year medical student at the University of Melbourne, with a strong interest in neurology and paediatric neurology. Prior to medical school, he completed a Bachelor of Science, where he developed a foundation in biomedical research. His academic interests include neuromuscular disorders, gene therapy, and paediatric neurology. Patrick Reardon passionate about translational medicine and hopes to contribute to advancing neurological treatment strategies.

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