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Research Center on Duchenne Muscular Dystrophy (DMD)

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News and Views

Duchenne Muscular Dystrophy (DMD) is an X-linked recessive muscular dystrophy affecting roughly 1 in 3500 boys, which is caused by mutations in the dystrophin gene on the short arm of chromosome Xp21.2 committed to encode “Dystrophin” protein, the key connector between cytoskeleton of a muscle fiber to the surrounding extracellular matrix through cell membrane, causing gradual loss of muscle tissue and function which eventually leads to wheelchair dependency at approximately the age of 12 years, requirement for assisted ventilation at approximately the age of 20 years and eventually premature death. Currently, there is no cure for DMD, but improvements in integrative treatment can slow down the disease progression and thereby extend the life expectancy of DMD patients. Patients with DMD have different forms of mutations at varying positions of the protein, resulting in the production of functionally compromised dystrophin ORF. The hallmark of DMD is the lack of presence of cellular dystrophin, the cementing protein linking actin cytoskeleton to muscle cell membrane. However, the selective absence of dystrophin can be reversed by the overexpression of utrophin, a close homolog of dystrophin. Mutations in the gene causing a disruption of the open reading frame or introduction of a premature stop codon lead to a complete absence of a functional dystrophin protein.

One of the main strategies of the current research towards the treatment of DMD is to restore the expression and function of the dystrophin gene. Despite its severity in terms of systemic muscle impairment culminating into multi organ failure and death, this disease is so far neglected due to lack of proper theranostic tools for in-time diagnosis and treatment. This project aims to address major issues through development therapeutic leads and its formulation for the treatment of DMD patients. Recently, DST-SERB India has sanctioned grant for establishment of First Government of India Funded Centre on Duchenne Muscular Dystrophy in India at IIT Jodhpur. This project will be implemented in collaboration with AIIMS Jodhpur and DART Bangalore. Further, IIT Jodhpur has planned a future roadmap for the supporting of DMD patients from state of Rajasthan as well as other parts of country.

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