
Fukutin-Related Protein: From Pathology to Treatments

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Abstract

Fukutin-related protein (FKRP) is a glycosyltransferase involved in the functional glycosylation of α-dystroglycan (DG), a key component in the link between the cytoskeleton and the extracellular matrix (ECM). Mutations in FKRP lead to dystroglycanopathies with broad severity, including limb-girdle and congenital muscular dystrophy. Studies over the past 5 years have elucidated the function of FKRP, which has expanded the number of therapeutic opportunities for patients carrying FKRP mutations. These include small molecules, gene delivery, and cell therapy. Here we summarize recent findings on the function of FKRP and describe available models for studying diseases and testing therapeutics. Lastly, we highlight preclinical studies that hold potential for the treatment of FKRP-associated dystroglycanopathies.

Keywords: congenital muscular dystrophy; dystroglycanopathy; glycosylation; limb-girdle; muscular dystrophy; α-dystroglycan.

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