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Functional protein networks unifying limb girdle muscular dystrophy

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Limb Girdle Muscular Dystrophy (LGMD) is a rare progressive heterogeneous disorder that can be caused by mutations in at least 21 different genes. These genes are often widely expressed and encode proteins with highly differing functions. And yet mutations in all of them give rise to a similar clinical presentation: adult onset muscle weakness, with muscles of the pelvic and shoulder girdle as predominantly affected muscle groups.

This thesis explores a potential molecular mechanism that unifies the different genetic defects, which individually can cause a Limb Girdle Muscular Dystrophy, with a focus on LGMD2A and 2B.

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