New gene linked to muscular dystrophy

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Muscular dystrophy, a group of inherited diseases characterized by progressive skeletal muscle weakness, can be caused by mutations in any one of a number of genes. Another gene can now be added to this list, as Yukiko Hayashi and colleagues, at the National Center of Neurology and Psychiatry, Japan, have now identified mutations in a gene not previously linked to muscular dystrophy as causative of a form of the disease in five nonconsanguineous Japanese patients.

Mutations in the caveolin-3 gene have previously been linked to muscular dystrophy.

In this study, the authors identified five nonconsanguineous Japanese patients with muscular dystrophy and degeneration of their fat tissue (a condition known as lipodystrophy) whose muscles were deficient in caveolin-3 protein in the absence of mutations in their caveolin-3 gene. Detailed genetic analysis revealed that these individuals had mutations in their PTRF gene, which is responsible for making a protein thought to influence caveolin protein stabilization.

Further investigation confirmed this as a function for the PTRF protein, as the mutated forms of the PTRF gene generated mutant PTRF proteins that could not localize correctly or associate with caveolin proteins. The authors therefore conclude that disease in the five patients studied is likely to be a result of caveolin deficiencies secondary to the PTRF gene mutations.

More information: Human PTRF mutations cause secondary deficiency of caveolins resulting in muscular dystrophy with generalized lipodystrophy, *Journal of Clinical Investigation*.

Source: Journal of Clinical Investigation