

Early-onset lumbar disc degeneration-associated mutation identified

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Lumbar disc degeneration (LDD) is characterized by pain in the lumbar region of the spine as a result of a compromised disc. LDD is fairly common and thought to be the result of both environmental and genetic risk factors; however, the genetic factors that promote LDD are largely unknown.

In this issue of the *Journal of Clinical Investigation*, Danny Chan and colleagues at the University of Hong Kong found mutations that reduced production of carbohydrate sulfotransferase 3 (CHST3) were associated with early-onset LDD. Mutations in families with LDD were mapped to the 3' untranslated region of the *CHST3* gene, which contained a microRNA binding site.

The authors determined that LDD-associated [mutations](#) in the 3' untranslated region enhanced microRNA binding, resulting in decreased *CHST3* expression. Furthermore, patients with early-onset LDD had decreased *CHST3* mRNA levels in intervertebral discs.

This study indicates that LDD development can be predicted by decreased *CHST3* expression.

More information: Lumbar disc degeneration is linked to a carbohydrate sulfotransferase 3 variant, *J Clin Invest*. [DOI: 10.1172/JCI69277](#)

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