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## Mouse models of nesprin-related diseases.

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### Abstract

Nesprins (nuclear envelope spectrin repeat proteins) are a family of multi-isomeric scaffolding proteins. Nesprins form the Linker of Nucleoskeleton-and-Cytoskeleton (LINC) complex with SUN (Sad1p/UNC84) domain-containing proteins at the nuclear envelope, in association with lamin A/C and emerin, linking the nucleoskeleton to the cytoskeleton. The LINC complex serves as both a physical linker between the nuclear lamina and the cytoskeleton and a mechanosensor. The LINC complex has a broad range of functions and is involved in maintaining nuclear architecture, nuclear positioning and migration, and also modulating gene expression. Over 80 disease-related variants have been identified in *SYNE-1/2* (nesprin-1/2) genes, which result in muscular or central nervous system disorders including autosomal dominant Emery-Dreifuss muscular dystrophy, dilated cardiomyopathy and autosomal recessive cerebellar ataxia type 1. To date, 17 different nesprin mouse lines have been established to mimic these nesprin-related human diseases, which have provided valuable insights into the roles of nesprin and its scaffold LINC complex in a tissue-specific manner. In this review, we summarise the existing nesprin mouse models, compare their phenotypes and discuss the potential mechanisms underlying nesprin-associated diseases.

**KEYWORDS:** DCM; EDMD; mouse models; nesprinPMID: 29784648 DOI: [10.1042/BST20180085](https://doi.org/10.1042/BST20180085)

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