

BACKGROUND

The *LMNA* gene encodes nuclear lamin A and nuclear lamin C, intermediate filament proteins that are components of the nuclear lamina.¹ These A-type lamins are expressed in virtually all differentiated somatic cells. Lamin A (664 amino acids) and lamin C (574 amino acids) arise via alternative splicing of pre-mRNA encoded by exon 10. Lamins A and C are identical for the first 566 amino acids. Lamin A is synthesized as a precursor, prelamin A, which has 98 unique C-terminal amino acids. Prelamin A protein has the amino acids CSIM (Cys-Ser-Ile-Met) at its C-terminus, which triggers a series of enzymatic reactions that lead to farnesylation and carboxylmethylation of the cysteine and endoproteolytic cleavage of the SIM peptide. The farnesylated prelamin A is then recognized by the endoprotease ZMSTE24, which cleaves 15 amino acids up from the cysteine residue at the C-terminus to yield lamin A. By contrast, lamin C has six unique C-terminal amino acids and is not post-translationally modified by farnesylation. Two other genes in the mammalian genome, *LMNB1* and *LMNB2*, encode lamins B1 and B2, respectively. Lamins A and C are widely expressed in the majority of differentiated somatic cells but are lacking from early embryos and from some undifferentiated cells, whereas lamins B1 and B2 are expressed in all or most somatic cells. There are little data and no systemic studies on the differences in the relative amount of lamins A, C, B1 and B2 expression. Lamins are intermediate filament proteins. Like all intermediate filament proteins, lamins contain a highly conserved α -helical core of approximately 350 amino acid residues flanked by globular N-terminal head and C-terminal tail domains. Lamins polymerize within the nucleus to form the nuclear lamina, a meshwork of intermediate filaments. The nuclear lamina is attached to the inner nuclear membrane by the binding of lamins to integral proteins of the membrane, among other factors. One function of the lamina is to provide structural support to the nucleus, and many have argued that defects in this function can lead to disease. Indeed, deficiency of A-type lamins leads to defective nuclear mechanics and defective mechanotransduction in cultured fibroblasts. Nuclear lamins have also been implicated in processes such as chromatin organization, gene regulation, DNA replication and RNA splicing.² Mutations in the *LMNA* gene cause a diverse range of diseases, called laminopathies, that selectively affect different tissues and organ systems. The most prevalent laminopathy is cardiomyopathy with or without different types of skeletal muscular dystrophy. *LMNA* cardiomyopathy has an aggressive clinical course with higher rates of deadly arrhythmias and heart failure than most other heart diseases.³

References:

1. Shackleton, S. et al: Nature Genet. 24:153-6, 2000
2. Dechat, T. et al: Genes Dev. 22:832-53, 2008
3. Vytopil, M. et al: J. Med. Genet. 40:e132, 2003

TECHNICAL INFORMATION

Source:

LMNA Antibody is a mouse monoclonal antibody raised against recombinant human LMNA fragments expressed in *E. coli*.

Specificity and Sensitivity:

This antibody detects LMNA proteins in various cell lysate.

Storage Buffer: PBS and 30% glycerol

Storage:

Store at -20°C for at least one year. Store at 4°C for frequent use. Avoid repeated freeze-thaw cycles.

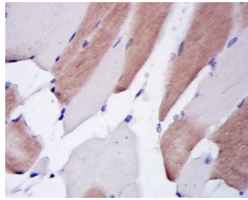
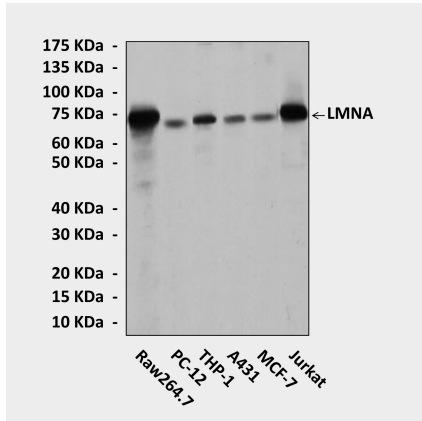
APPLICATIONS

Application:	*Dilution:
WB	1:1000
IP	n/d
IHC (Paraffin)	1:50-200
ICC	n/d
FACS	n/d

**Optimal dilutions must be determined by end user.*



QUALITY CONTROL DATA



Top: Western blot detection of LMNA proteins in various cell lysates using LMNA Antibody. **Bottom:** This antibody stains paraffin-embedded human striated muscle tissue in IHC analysis.

