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Recombinant Human Prelamin A R527H

REP0048
100µg

Description

Prelamin A is the precursor of mature lamin A, a structural protein of the nuclear lamina, and it is encoded by LMNA gene. Prelamin A carrying the R527H mutation is found in subjects affected by Mandibuloacral Dysplasia type A [MADA; OMIM no. 248370].

MADA is a rare and complex disease characterized by postnatal growth retardation, craniofacial anomalies, skeletal malformations, mottled cutaneous pigmentation, partial lipodystrophy (type A pattern), and insulin resistance. MADA patients seem to be genetically homogeneous, since they show the same mutation (R527H) in the LMNA gene. In contrast, patients with generalized loss of subcutaneous fat involving the face, trunk, and extremities (type B pattern) carry mutations in the ZMPSTE24 gene (MADB; OMIM no. 608612).

MADA is a specific genetic entity belonging to a class of genetic disorders called "laminopathies". The R527H lamin A/C mutation produces accumulation of unprocessed prelamins A, altered distribution of the lamin B receptor (LBR), and destabilization of two heterochromatin-associated proteins, histone H3 methylated at lysine 9 (Me9H3) and heterochromatin protein-1β (HP1β).

Product type

Protein

Peptide

Recombinant full-length protein: Human Prelamin A – Accession number: **NP_733821** (1 to 664) with N-terminal His tag and DDDDK tag carrying the R527H mutation

Sequence

MAHHHHHHVGTGSDDDDDKSPDMETPSQRRATRSGAQASSTPLSPTRITRLQEKEDLQELNDR LAVYIDRVHSLETENAGLRLRI
TESEEVVSREVSGIKAAAYEALGDARKTLDSVAKERARLQLELSKVREEFKELKARNTKKEGDLIAAQARLKDLEALLNSKEAALSTA
LSEKRTLEGELHDLRGQVAKLEAALGEAKKQLQDEMLRRVDAENRLQTMKEELDFQKNIYSEELRETKRRHETRLVEIDNGKQREF
ESRLADALQELRAQHEDQVEYKKELEKTYSAKLDNARQSAERNSNLVGAHEELQQSRIRIDLSAQLSQLQKQLAAKEAKLRDL
EDSLARERDTSRRLAEEKEREMAEMRARMQQQLDEYQELLDIKLALDMEIHAYRKLLEGEERLRRLSPSPQRSRGRASSHSST
QGGGVSVTKKRKLESTESRSSFQHARTSGRVAVEEVDEEGKFVRLRNKSNEDQSMGNWQIKRQNGDPLLTYRFPPKFTLKAGQ
VVTIWAAGAGATHSPPTDLVWKAQNTWGCNSLHTALINSTGEEVAMRKLVRSVTVVDEDEDEDGDDLLHHHHGSHCSSSGDP
AEYNLRSRTVLCGTCGQPADKASASGSGAQVGGPISSGSSASVTVTRSYRSYRSGGSGGSGFDNLVTRSYLLGNSSPRTQSPONC
SIM

Expression system

Escherichia coli

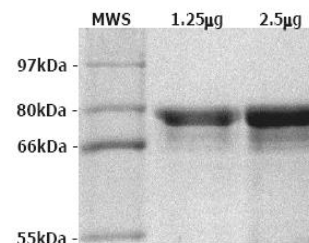
Tested by

SDS Page, Western Blotting

Purity

>90% pure estimated by SDS-PAGE (EU Ph. 5.0 § 2.5.31)

Purified recombinant full-length prelamins A R527H separated by SDS-PAGE (8% polyacrylamide stained with Coomassie Blue).



Form

Liquid

Storage Buffer

20mM phosphate buffer pH 7.0, 0.5M NaCl, 1mM DTT, 1.5mM EDTA, 25%(v/v) glycerol.

Storage instructions

Shipped on dry ice. Upon delivery aliquot and store at -20°C. Avoid freeze / thaw cycles.