



and recover completely. Molecular determinant of the late arterial morphogenesis, stabilizing arterial structure by regulating proliferation and organization of vascular smooth muscle.

#### 組織特異性

Expressed within the outer myometrial smooth muscle and throughout the arteriolar tree of uterus (at protein level). Also expressed in the large arteries, lung and skin.

#### 関連疾患

Defects in ELN are the cause of cutis laxa, autosomal dominant, type 1 (ADCL1) [MIM:123700]. A connective tissue disorder characterized by loose, hyperextensible skin with decreased resilience and elasticity leading to a premature aged appearance. Face, hands, feet, joints, and torso may be differentially affected. Additional variable clinical features are gastrointestinal diverticula, hernia, and genital prolapse. Rare manifestations are pulmonary artery stenosis, aortic aneurysm, bronchiectasis, and emphysema.

Defects in ELN are the cause of supravalvular aortic stenosis (SVAS) [MIM:185500]. SVAS is a congenital narrowing of the ascending aorta which can occur sporadically, as an autosomal dominant condition, or as one component of Williams-Beuren syndrome.

Note=ELN is located in the Williams-Beuren syndrome (WBS) critical region. WBS results from a hemizygous deletion of several genes on chromosome 7q11.23, thought to arise as a consequence of unequal crossing over between highly homologous low-copy repeat sequences flanking the deleted region. Haploinsufficiency of ELN may be the cause of certain cardiovascular and musculo-skeletal abnormalities observed in the disease.

#### 配列類似性

Belongs to the elastin family.

#### 翻訳後修飾

Elastin is formed through the cross-linking of its soluble precursor tropoelastin. Cross-linking is initiated through the action of lysyl oxidase on exposed lysines to form allysine. Subsequent spontaneous condensation reactions with other allysine or unmodified lysine residues result in various bi-, tri-, and tetrafunctional cross-links. The most abundant cross-links in mature elastin fibers are lysinonorleucine, allysine aldol, desmosine, and isodesmosine.

Hydroxylation on proline residues within the sequence motif, GXPG, is most likely 4-hydroxy as this fits the requirement for 4-hydroxylation in vertebrates.

#### 細胞内局在

Secreted > extracellular space > extracellular matrix. Extracellular matrix of elastic fibers.

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