

Supplemental Table 2. Correlation of gene expression domains in cultured fibroblasts with phenotypes in genetic diseases. For detailed descriptions of mutant alleles and clinical features, please see Online Mendelian Inheritance in Man and references therein.

<u>Diseases</u>	<u>Gene/ Protein</u>	<u>Site of Overexpression</u>	<u>Clinical features</u>
Bethem myopathy	COL6A1	thigh, abdomen, back	Proximal limb-girdle myopathy with joint contractures
Conradi-Hunerman syndrome	emopamil binding protein	arm, abdomen	Whorled ichthyosis, chondrodysplasia punctata, rhizomelic shortening of limbs
Ehler-Danlos I	Collagen type V COL1A1	fetal skin	Hyperextensile skin. Loose joints. Fragile skin, easy bruising and heals with "cigarette paper" scars. COL5A1 is also mutated in EDS2 (milder phenotype); COL1A1 also mutated in EDS7.
Ehler-Danlos V	lysine oxidase	cutaneous	Similar to type I but less severe. X-linked.
Ehler-Danlos VI	lysine hydroxylase	fetal skin	Similar to type I plus kyphoscoliosis or ocular complications such as retinal detachment or keratoconus.
Ehler-Danlos X	Fibronectin	cutaneous	Similar to type I plus defect in collagen-induced platelet aggregation.
hand-foot-genital syndrome	HOXA13	toe, foreskin	Hypoplastic distal phalanges, thenar eminence. Hypospadias in males, bifid uterus in females and anomalies in urogenital system.
Marfan's syndrome	Fibrillin	cutaneous	Striae distensae. Increased height and limb length. Loose joints; mitral valve prolapse and risk of thoracic aneurysm.
osteogenesis imperfecta	COL1A1	fetal skin	Thin translucent skin, multiple fractures, blue sclera, poor dentition and otosclerosis.
pycnodysostosis	cathepsin K	cutaneous and gum	Short stature, brachycephaly, short fingers, osteosclerosis and open cranial sutures.
striate palmo-plantar keratoderma	desmoplakin I	toe	Thick hyperkeratotic linear papules on palms and soles.
ulnar-mammary syndrome	TBX3	foreskin, gum	defect in apocrine differentiation. Hypogenitalism, dental abnormalities. Postaxial limb defects
X-linked ichthyosis	arylsulfatase C	cutaneous	Diffuse ichthyosis