Cutis Laxa

Gene panel

Gene panel information

Gene panel	**Cutis Laxa**
Version	1
Total genes	19
Activation date	Wednesday 21 december 2022
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ALDH18A1	99.91 %	138250	Spastic paraplegia 9A, autosomal dominant, 601162 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IIIA, 219150 (3), Autosomal recessive; Spastic paraplegia 9B, autosomal recessive, 616586 (3), Autosomal recessive; Cutis laxa, autosomal dominant 3, 616603 (3), Autosomal dominant
ATP6V0A2	99.77 %	611716	Wrinkly skin syndrome, 278250 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive
ATP6V1A	99.41 %	607027	Cutis laxa, autosomal recessive, type IID, 617403 (3), Autosomal recessive; Developmental and epileptic encephalopathy 93, 618012 (3), Autosomal dominant
ATP6V1E1	99.83 %	108746	Cutis laxa, autosomal recessive, type IIC, 617402 (3), Autosomal recessive
ATP7A	99.79 %	300011	Occipital horn syndrome, 304150 (3), X-linked recessive; Neuronopathy, distal hereditary motor, X-linked, 300489 (3), X-linked recessive; Menkes disease, 309400 (3), X-linked recessive
COG7	98.84 %	606978	Congenital disorder of glycosylation, type IIe, 608779 (3), Autosomal recessive
EFEMP1	99.4 %	601548	Doyne honeycomb degeneration of retina, 126600 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type ID, 620780 (3), Autosomal recessive; Glaucoma 1, open angle, H, 611276 (3), Autosomal dominant
EFEMP2	99.7 %	604633	Cutis laxa, autosomal recessive, type IB, 614437 (3), Autosomal recessive
ELN	98.97 %	130160	Cutis laxa, autosomal dominant, 123700 (3), Autosomal dominant; Supravalvar aortic stenosis, 185500 (3), Autosomal dominant
EMILIN1	99.81 %	130660	Neuronopathy, distal hereditary motor, autosomal dominant 10, 620080 (3), Autosomal dominant; Arterial tortuosity-bone fragility syndrome, 620908 (3), Autosomal recessive
FBLN5	99.73 %	604580	Cutis laxa, autosomal recessive, type IA, 219100 (3), Autosomal recessive; Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764 (3), Autosomal dominant; Macular degeneration, age-related, 3, 608895 (3), Autosomal dominant; ?Cutis laxa, autosomal dominant 2, 614434 (3), Autosomal dominant
GORAB	99.51 %	607983	Geroderma osteodysplasticum, 231070 (3), Autosomal recessive
LOX	99.8 %	153455	Aortic aneurysm, familial thoracic 10, 617168 (3), Autosomal dominant
LTBP1	99.63 %	150390	Cutis laxa, autosomal recessive, type IIE, 619451 (3), Autosomal recessive
LTBP4	99.61 %	604710	Cutis laxa, autosomal recessive, type IC, 613177 (3), Autosomal recessive
NAA10	99.94 %	300013	Microphthalmia, syndromic 1, 309800 (3), X-linked; Ogden syndrome, 300855 (3), X-linked dominant, X-linked recessive
PYCR1	99.75 %	179035	Cutis laxa, autosomal recessive, type IIIB, 614438 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIB, 612940 (3), Autosomal recessive
RIN2	99.88 %	610222	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075 (3), Autosomal recessive
TALDO1	99.87 %	602063	Transaldolase deficiency, 606003 (3), Autosomal recessive

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2025-11-12**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

Possible phenotype mapping keys

(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known

(2) the disorder has been placed on the map by linkage; no mutation has been found

(3) the molecular basis for the disorder is known; a mutation has been found in the gene

(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.