Cutis laxa en gerelateerde aandoeningen v3.0 (13 genen)

Amsterdam UMC
Universitair Medische Centra

Gene	% covered > 30x	Associated phenotype description and OMIM ID
ALDH18A1	>99	Cutis laxa autosomal dominant, 3[illegible]1660 Cutis laxa autosomal recessive type IIIA, 219150 Spastic paraplegia 9A autosomal dominant, [illegible]0116 Spastic paraplegia 9B autosomal recessive, [illegible]1658
ATP6V0A2	>99	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP6V1A	>99	Cutis laxa, autosomal recessive, type IID, 617403 Epileptic encephalopathy, infantile or early childhood, 3, 618012
ATP6V1E1	>99	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7A	>99	Menkes disease, [illegible]0940 Occipital horn syndrome, [illegible]0415 Spinal muscular atrophy distal X-linked, 3[illegible]0048
EFEMP2 (FBLN4)	>99	Cutis laxa, autosomal recessive, type IB, 614437
ELN	>99	Cutis laxa, AD, 123700 Supravalvar aortic stenosis, 185500
FBLN5	>99	Cutis laxa autosomal dominant, 2[illegible]1443 Cutis laxa autosomal recessive type IA, [illegible]1910 Macular degeneration age-related, 3[illegible]0889 Neuropathy hereditary with or without age-related macular degeneration, [illegible]0889
GORAB	>99	Geroderma osteodysplasticum, 231070
LTBP4	>99	Cutis laxa, autosomal recessive, type IC, 613177
PYCR1	>99	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
RIN2	>99	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
SLC2A10	>99	Arterial tortuosity syndrome, 208050

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
OMIM release used for OMIM disease identifiers and descriptions : June 30th, 2015
"No OMIM phenotype" signifies a gene without a current OMIM association
OMIM phenotype descriptions between {} signify risk factors