Lens luxatie en gerelateerde syndromen v3.0
(10 genen, incl. 4 core-NL genen)

Amsterdam UMC
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Gene	% covered >30x	Associated phenotype description and OMIM ID
ADAMTS10	>99	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	>99	Weill-Marchesani-like syndrome, 613195
ADAMTSL4	>99	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ASPH		Traboulsi syndrome, 601552
CBS	>99	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
COL18A1	>99	Glaucoma, primary closed-angle, 618880 Knobloch syndrome, type 1, 267750
CPAMD8	>99	Anterior segment dysgenesis 8, 617319
FBN1	>99	Acromicric dysplasia, 10237 Aortic aneurysm ascending and dissection Ectopia lentis familial, 12960 Geleophysic dysplasia, 261418 Marfan lipodystrophy syndrome, 51691 Marfan syndrome, 15470 MASS syndrome, 50430 Stiff skin syndrome, 18490 Weill-Marchesani syndrome 2 dominant, 60832
LTBP2	>99	Glaucoma 3 primary congenital D, 61308 Microspherophakia and/or megalocornea with ectopia lentis and with or without secondary glaucoma, 25175 Weill-Marchesani syndrome 3 recessive, 61481
P3H2	>99	Myopia, high, with cataract and vitreoretinal degeneration, 614292

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