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Supplemental Data

Homozygous Mutations in *ADAMTS10* and *ADAMTS17* Cause Lenticular Myopia, Ectopia Lentis, Glaucoma, Spherophakia, and Short Stature

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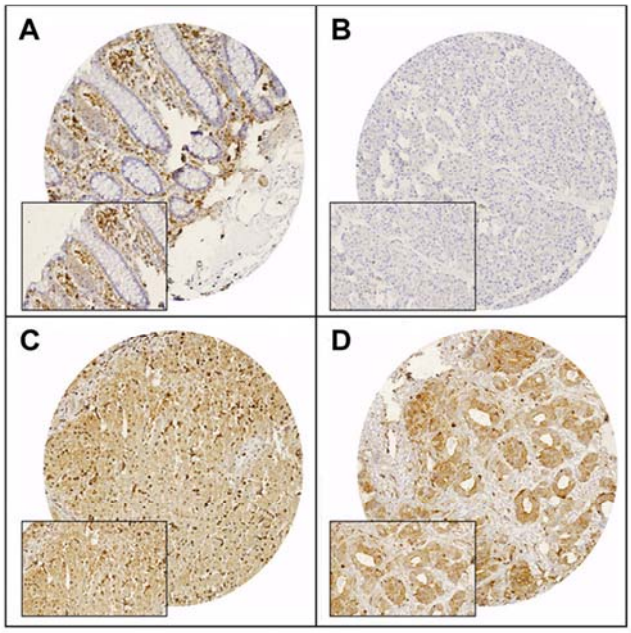
Figure S1: Immunohistochemical Studies of ADAMTS17. **I)** Western blot assay using ADAMTS17 monoclonal antibody on three different cell lines showing the ADAMTS17 estimated band size of 121kDa. **II)** Tissue microarray based Immunohistochemical studies of ADAMTS17 in human tissues **(A**) weak cytoplasmic staining, 1+ in normal colorectal mucosa, **(B**) no staining, 0 in pancreatic tissue **(C)** moderate cytoplasmic (2+)as well as membranous staining, (1+) in liver (**D**) strong staining, 3+ in the cytoplasmic and membranous compartment in epithelial ovarian carcinoma. 20 X/0.70 objective on an Olympus BX 51 microscope (Olympus America Inc, Center Valley, PA, USA) with the inset showing a 40X/ 0.85 aperture magnified view of the same. **III)** IHC on mouse E14.5 eye section using ADAMTS17 monoclonal mouse antibody, the ciliary body is outlined in box.

I)

II)



III)

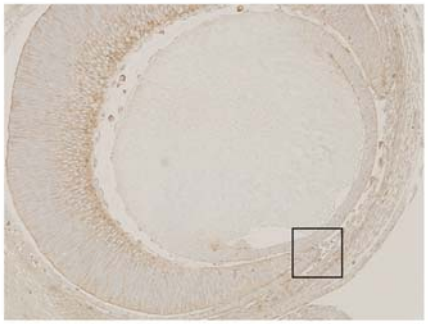


Table S1: Primers and PCR Conditions for *FBN1* Gene.

Gene/Exon	Primer Sequence 5' – 3'	Product Size	Annealing Temperature °C/Cycles
FBN1X2F FBN1X2R	AGCAGCGCCTGGAGAAG GGTGGGGACTAAACAACCCT	467bp	50/30 +10% DMSO
FBN1X3F FBN1X3R	GCTTTATCTGTCTGCCAGGATT AAGCTGGAAGGCTGTACTATCAA	333bp	56/30
FBN1X4F FBN1X4R	ACACCTGAGGCTTAAGGTCATA GAGAAGGCAGATGTTCTAGTGAAA	368bp	56/30
FBN1X5F FBN1X5R	GCCGGTAAATGATTTTACACAC CCAATTGGAAATTTGGTGT	470bp	52/30
FBN1X6F FBN1X6R	GTCCAAAACAAACACCAGGG TAGGGACCTTCCCAATGACA	365bp	56/30
FBN1X7F FBN1X7R	TGCATGATTCTGTCCCTGAA TGGATATCATGCAGTCAGCG	460bp	56/30
FBN1X8F FBN1X8R	TGATGGACAAATAACTCACCGA AGCACAAAGGCATATTATTACTGTTA	415bp	56/30
FBN1X9F FBN1X9R	ACTGTTCTCCATGCGGTTTT TGCCTCTGTTCTTTAAGAGGG	438bp	52/30
FBN1X10F FBN1X10R	GGGGCAGAGGTGTGAGTTAAT TTCCCTGGACGTCATCTCTT	425bp	52/30
FBN1X11F FBN1X11R	GGTTGCTGCTGCAGGTATTA TCTATGCAATAGGAAAATTGAGACA	551bp	56/30
FBN1X12F FBN1X12R	GCAATGTGATCTCTGTTGGC CCTCCCAACACCTGGATG	514bp	56/30
FBN1X13F FBN1X13R	TGGCTAATGACATATTGTAGTGGA CAGAATGACAATAGGTGGAACTAAG	435bp	56/30
FBN1X14F FBN1X14R	TGTCTTTTCAACTCTGATAACCCT TCATGGAAAATTAGGCTTCCT	459bp	56/30
FBN1X15F FBN1X15R	GATCTTATTTGGATGAAAGTTAGCC TCCCAAACCAAAATTCAAGG	391bp	56/30
FBN1X16F FBN1X16R	TGAGCTTTTGTTGCTGATGC TGGTTTGTTGCTCTGCAAAT	384bp	56/30
FBN1X17F FBN1X17R	CCCCGATATCAGAGGCATT AAGACCCCAAGAAGGCACAT	416bp	50/35+10% DMSO
FBN1X18F FBN1X18R	GGGAGATACCTCCTTTTCCG GATGGCCAGAGAGGGAGTC	310bp	56/30
FBN1X19F FBN1X19R	TGTTCTCTTTCCAGTGAAATCG AACATCCGAAGTATAAAGTGTCCA	393bp	56/30
FBN1X20F FBN1X20R	TGCTACAGGAGTTTTGCCTTT CACAGAATTTCAACTAAACTGGCA	386bp	56/30
FBN1X21F FBN1X21R	AGCCCAGCTTTACTGTGTGG CAAAAATAGCATTTGAAACAAGGA	377bp	56/30
FBN1X22F FBN1X22R	TCCTTGTTTCAAATGCTATTTTTG CCCATTCAGCAATATGTTCG	375bp	56/30
FBN1X23F FBN1X23R	CCTATGCAATCGCAGAAACA CCTGTCCTTTGGACATAGCG	426bp	56/30
FBN1X24F FBN1X24R	TTGCTATTCAGGCACCCTAGA CATGCTGAATTTTGGAGTGTG	413bp	50/30
FBN1X25F FBN1X25R	GGCAAGGATACTTACCCCAGA CCAGGACTTCTTGGACCAAAC	588bp	56/30
FBN1X26F FBN1X26R	TGGATCATCACACATTCTTGG CAAGTGGCAGCAAATGAGTC	385bp	56/30
FBN1X27F FBN1X27R	AGCCTTAAGGGCCAGGAGAG CCTCAGTCTCCCTCTGTTGC	389bp	56/30
FBN1X28F FBN1X28R	CCAGAGAGTTGTCCCACCTT CATTGATGTCTGTCGGGAAA	374bp	56/30
FBN1X29F FBN1X29R	ATCTCCGCGTGTATCGGTAA AGCGATGAAAACAAAACTCAGA	390bp	56/30
FBN1X30F FBN1X30R	GATCCCACCATGAGGGTAGA GCAGGCAATTTGAACTTCATT	470bp	56/30
FBN1X31F FBN1X31R	TTATGCAAAGCTTCATTTGGA TGTGCCTCTAAATATCCTTACTTTTC	396bp	56/30
FBN1X32F	AGCAAATGTAAATTGGCCTCT	441bp	50/35

FBN1X32R	AATTTCAAAGAAGTGGAAGCTAAA		
FBN1X33F FBN1X33R	TGGGAAGTTTGAAGGCAAGT ATCATTATTAACAGAAAGGGTGGT	356bp	56/30
FBN1X34F FBN1X34R	GAGGTTGCTTATCAAGGACTGC GAGCTCTAGTGCCAAATGATGA	389bp	56/30
FBN1X35F FBN1X35R	TGTCAAATTGAAGTTATGCCAAA AGCTGGAAACCTAATCCCTCT	400bp	50/35
FBN1X36F FBN1X36R	GCCCAGATTGGTGTTAGATACTC TGAGAAAAGGTATCTGTGATTCTTAAT	393bp	56/30
FBN1X37F FBN1X37R	TCTGAAGTGGAAGACTGCATTT TTCCCTATGGAAAAGATATCTGAA	431bp	50/35+ 10%DMSO
FBN1X38F FBN1X38R	TCGGTAGTGTATAATGTTCATGGG CTACAGGGCTGAGAGGACTGA	435bp	56/30
FBN1X39-40F FBN1X39-40R	TCAGACGGGCAGAGTAACAA CCTGGCTATGTTCGTGTTTAGA	556bp	56/30
FBN1X41F FBN1X41R	GAGGACACGGATGAATGAAA AGGGATGCCAGCACTCAG	393bp	56/30
FBN1X42F FBN1X42R	TTTTGTGTCCTTATCAAAGTCAAG TGACCAGCACCAACTGTGAA	452bp	56/30
FBN1X43F FBN1X43R	ACAGGCAAGATTATATCTGTAGGAA CAGGGTGTTTGCACAGTTTG	406bp	56/30
FBN1X44F FBN1X44R	GTCCCTATTGCCATCACCAC TCCACACCATGCCCTTTACT	411bp	56/30
FBN1X45F FBN1X45R	CGAAGGACATCTTGGTTGCT TTTTGACAATATCCCTTTTTCCA	470bp	56/30
FBN1X46F FBN1X46R	TGCCTGGTAAATGAGAGTAAGTT TCACTGCTGCATATCTGTCTG	440bp	56/30
FBN1X47F FBN1X47R	TCAAGAAGTTCTCAGCCTATGGA TGTATTTGACAAGTCCCATAACCA	386bp	56/30
FBN1X48F FBN1X48R	AAGCCAGCAAAGGCCACTA AAAACAAGCCTTTATAGGGAGGA	397bp	50/35+10% DMSO
FBN1X49F FBN1X49R	TTTTCTCCATGGTGGAATTTT CTTGCCAGAAGGATGAGACC	415bp	50/35+10% DMSO
FBN1X50F FBN1X50R	TCCACAGTGTTATGGTACAGAAA GGGTGAATGAAAACTCTCCATC	396bp	50/35+10% DMSO
FBN1X51F FBN1X51R	GGAGTATTGCTGTGGTCCTGA GAAAATAACTACCAAGCTCCTGAGA	441bp	50/35
FBN1X52F FBN1X52R	CATCATTTATCAATATGCACAGCA GCACTTAATTTTCCAAGATAGATGG	280bp	52/30
FBN1X53F FBN1X53R	TTTGTCCCTTCATTTAGATAGCAA AACTTATTTCAGTGCCATCTTGG	362bp	50/35+ 10%DMSO
FBN1X54F FBN1X54R	TGGATGTCAAAGTGAGACTCTGT AATGATCAAATGGCCCATCA	387bp	52/30
FBN1X55F FBN1X55R	TGGGAGACCACTTGATCCAT CCGTATTGTCCACGGACTATTTA	390bp	52/30
FBN1X56F FBN1X56R	GAGCAGAAGGAAATACAGCCA CCTGACATGCGGTTAAGAGA	401bp	52/30
FBN1X57F FBN1X57R	AAAGGAACAAAGGGAGGGAA CGGCATCTCCAAAATATGAA	384bp	50/35+10% DMSO
FBN1X58F FBN1X58R	CTTGCTTCTTCTCACCAGG GGCAGCATGACCTCAAATGT	505bp	50/35+10% DMSO
FBN1X59F FBN1X59R	TCTTAGGCCCAAAATATAGTAACACA CGGGGAAAGAAAAAGGAAAC	400bp	52/30
FBN1X60F FBN1X60R	CCTTTTTCTTTCCCCGAAAC AAGCACCTCCTGCCTGTAGA	345bp	56/30
FBN1X61F FBN1X61R	CTCTCCTTGCCTTTTGCTGT AAAGGCCAAATAAGGCCAAC	437bp	52/30
FBN1X62F FBN1X62R	TCCTCTGCTGTTGGGATAAGA TCAAGCATGAAACAAAACACG	510bp	52/30
FBN1X63F FBN1X63R	TCTCCTTGGACTTAGCAGCAG GCTGTTTTGCAAAGACTGTTACTT	448bp	52/30
FBN1X64F FBN1X64R	GCCTGTCTTACCTTCCTGAGA CTTTTGCCAAGCTAACTGGA	502bp	52/30
FBN1X65F FBN1X65R	ATCCTCCAGTGGACAAGCAA GCTCAGGGAATGTCTGGAGT	433bp	50/35+10% DMSO
FBN1X66F FBN1X66R	TGCCAATTTAATACACTTGTGGTC TCACCTGTACCTTGCTTTGG	719bp	52/30

Table S2: Primers and PCR Conditions for *ADAMTS17* Gene

Gene/Exon	Primer Sequence 5' – 3'	Product Size	Annealing Temperature °C/Cycles
ADAMTS17X1F ADAMTS17X1R	TCCGCAGCGCTAATTACTTT ACCCGATTCCCGTACTCC	353bp	56/30 +10%DMSO
ADAMTS17X2F ADAMTS17X2R	AGGGAAGGGAAGGCAGC AGGTGGAAAGGCAGGGG	636bp	56/30 +10%DMSO
ADAMTS17X3F ADAMTS17X3R	CAAATCACAGCAAGGTGGTC TTCTCACTCATGTGGCTTCAGT	359bp	56/30
ADAMTS17X4F ADAMTS17X4R	CTGACCACTCTCTCGGGC ACCCAGCGTCTTCCTCACT	383bp	56/30
ADAMTS17X5F ADAMTS17X5R	TTTCTGTGTCCCAAGTTCCC AGAGAGTGACGGAGACTGGC	290bp	56/30
ADAMTS17X6F ADAMTS17X6R	GCCTCCACCTTAACGTCCTT GTGGCCCAAGGTCTGATTT	362bp	56/30
ADAMTS17X7F ADAMTS17X7R	TTGCAGGATGACTACTTGAGG GCTTGTTTGAGGACAGCTCC	237bp	56/30
ADAMTS17X8F ADAMTS17X8R	TTGATTGTGCATGACGTTGA CAGATGCAGCAAAGGCATAG	314bp	56/30
ADAMTS17X9F ADAMTS17X9R	AAGACTTGATTTACCTTAGCATGG TTCTCCACTTCACACTTGCTG	329bp	56/30
ADAMTS17X10F ADAMTS17X10R	GGAGGTGCCCTCTTCTCAGT GTGCTGAGTGTGAATGCCC	361bp	56/30
ADAMTS17X11F ADAMTS17X11R	ATTTGAAACAGTGGTTGGGC TGTTTCAGGGGCTCCTAAGT	304bp	56/30
ADAMTS17X12F ADAMTS17X12R	TGGCAGCACGGCATTTT CTTCTAATGCTCAGCGGGAG	342bp	56/30 with 10%DMSO
ADAMTS17X13F ADAMTS17X13R	CCCTTCCTCTCCTCCCC CTGGGTTGGTTTGGGAAAG	369bp	56/30
ADAMTS17X14F ADAMTS17X14R	CTTCCTCAGGGTTTCCCTCT CCTGAGACCTCTGCTCTACCC	315bp	56/30
ADAMTS17X15F ADAMTS17X15R	TATGCTGCACCTCATGGAAA TCCACTACCATCTAGCCCTTAAA	331bp	56/30
ADAMTS17X16F ADAMTS17X16R	CTGTGGAAGGGAGACCTCTG GAGGTCCCAGGCAGAAGTAA	361bp	56/30
ADAMTS17X17F ADAMTS17X17R	AAAGAGCTGGTTACCCCTCA ATGTCTCACACTCTGCGTGC	370bp	56/30
ADAMTS17X18F ADAMTS17X18R	GCACCCAAACATGCTTTCAT GCATAGAGCAGTACTGTGGCA	331bp	56/30 with 10%DMSO
ADAMTS17X19F ADAMTS17X19R	AGGGCAGCAATTCCAGGAT CAACTTCAAGCTGACCTGGG	415bp	56/30
ADAMTS17X20F ADAMTS17X20R	GCACCTGGCACATTGGA GCCTAGTTACGCTGGGACTG	357bp	56/30
ADAMTS17X21F ADAMTS17X21R	CCACTCAGGCTGTCCTGTCT TTTCTGGCTGAAAGAACCTCA	370bp	56/30
ADAMTS17X22F ADAMTS17X22R	AGTGCAAAGGGTTGACGTGT CCAAGTCCACGCTCATGTT	429bp	56/30

Table S3: Variants Identified in *ADAMTS17*.

Location	Nucleotide Change	Amino Acid Change	Identification Numbers [a]
Intron1	c.79+31 C>G	NA	rs2585224
Intron2	c.451-70 C>T	NA	rs11639041
Intron3	c.617-41 A>G	NA	rs7497712
Exon 4	c.647 C>T	S216L	rs7496668
Exon 4	c.756 C>T	A252	rs7496640
Intron4	c.789+8 C>T	NA	rs7496614
Intron 5	c.873+58 T>C	NA	rs4965297
Intron5	c.873+62 C>T	NA	rs4965296
Exon6	c.1017 C>T	A339	rs4965613
Intron6	c.1031+81 A>G	NA	rs4965295
Exon7	c.1053 G>A	K351	rs4369638
Intron9	c.1322+38 A>G	NA	rs8024690
Intron9	c.1323-11 G>A	NA	ss161151830
Exon10	c.1445 T>C	M482T	rs28567966
Exon12	c.1696 A>C	R566	rs12907333
Intron13	c.1889-44 G>A	NA	ss161151831
Exon14	c.1962 C>T	D654	rs61752832
Intron14	c.2016+87 T>G	NA	ss161151832
Intron15	c.2137+59 C>G	NA	ss161151833
Exon15	c.2112 C>T	G704	rs4965583
Intron15	c.2138-12 C>T	NA	rs28529328
Intron15	c.2138-40 C>G	NA	rs28587358
Inron16	c.2296-56 T>A	NA	rs4965569
Exon18	c.2472 C>T	I824	rs61752831
Intron18	c.2591+6 A>G	NA	rs8028251
Intron19	c.2796+96 G>A	NA	rs12900132
Intron20	c.2949+35 G>A	NA	rs7175995
Intron20	c.2949+42 A>G	NA	rs2573607
Exon22	c.3281 A>G	N1094S	rs2573652

NA: not applicable. [a]SNP database (dbSNP) identification numbers, reference SNP (rs) numbers for previously reported SNPs , submitter SNP (ss) numbers for the SNPs identified in this study.

Table S4: Localization of ADAMTS17 in Different Tissues Using IHC on TMA.

Tissue	Cytoplasm	Membrane
Colon	+1	0
Pancreas	0	0
Liver	+2	+1
Kidney	+2	0
Lung	+1	0
Heart	+1	0
Ovary	+1	0
Testis	+2	+3
Uterine smooth muscle	+1	0
Lymph node	+2	0
Spleen	+3	0
Placenta	+2	0
Thyroid	0	0
Ovarian cancer	+3	+3
Endometrial cancer	+3	+3
Colon cancer	+3	+2