

MUTATIONS IN THE GENE THAT ENCODES THE F-ACTIN BINDING PROTEIN ANILLIN CAUSE FSGS

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SUPPLEMENTARY MATERIALS

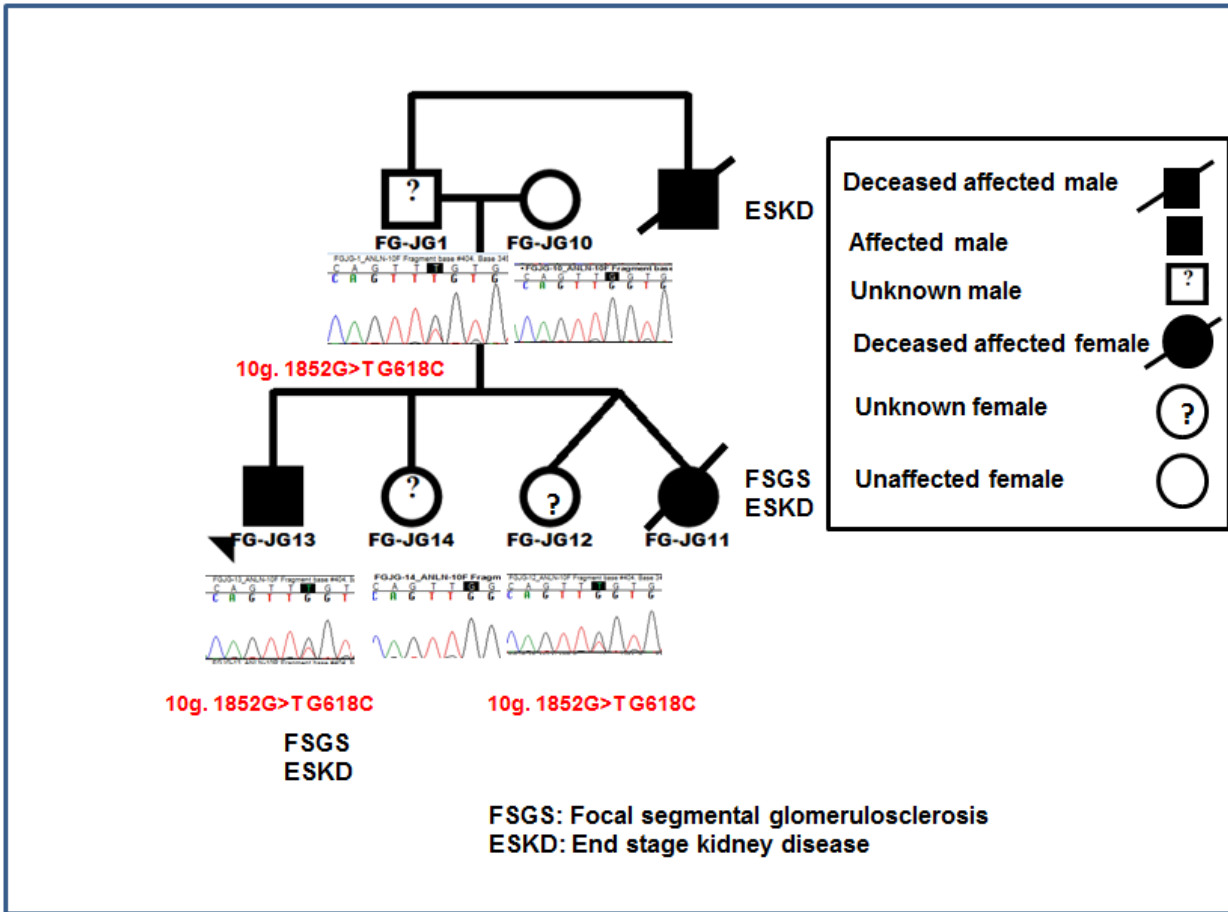


Figure S1: Pedigree and phenotype of second kindred with G618C mutation in *ANLN*.

Supplementary Table 1: Variants with Minor allele frequency <1% in chromosomes 2p, 5p and 7p locus

Locus	Variant ID/ Nucleotide Change	Present in in-house controls Yes/No	Gene/Type of variant
Chromosome 2p	2_26526439_T/ G>T	Yes	C2ORF39/ R527S
Chromosome 2p	2_27300747_A/ A>C	Yes	CAD/ P380H
Chromosome 2p	2_27583673_27583674_INS_A	No	GCKR/ Frame shift
Chromosome 2p	2_33598552_G/ G>A	No	RASGRP3/ N68S
Chromosome 2p	2_39094611_A/ G>A	Yes	SOS1/ P655L
Chromosome 5p	5_1090778_A/ G>A	No	NKD2/ V311I
Chromosome 7p	7_25142786_C/ C>T	No	C7ORF31/ H368R
Chromosome 7p	7_36417196_T/ C>T	No	ANLN/ R431C missense
Chromosome 7p	7_44527869_A/ G>A	Yes	NPC1L1/ P974S
Chromosome 7p	7_45684173_A/ G>A	No	ADCY1/ E596K

Supplementary Table 2: ANLN variants found in cohort of patients with FSGS

RS number	Exon/Intron	Nucleotide	Amino Acid Change
rs10215150	Intron 2	(-)130C>G	None
rs79086052	Intron 2	(-)153T>C	None
rs116131910	2	100A>G	M34V
rs138687537	2	143G>A	S48N
rs3735400	3	194C>G	S65W
rs34285732	3	434G>A	R145H
rs61737563	4	553A>C	None
rs62873663	4	554G>A	R185K
rs3735398	4	819A>G	None
1000 genomes	Intron 4	(+)25T>C	None
rs112558277	5	884_883insCTT	S295SS
rs61549495	5	886_885insTCT	296^295insS
rs3801315	Intron 5	(+)35T>C	None
rs11976268	6	1245C>T	None
rs113329122	Intron 7	(-)61G>A	None
rs10224902	8	1506T>C	None
1000 genomes	Intron 9	(-)27A>C	None
rs78667831	Intron 9	(+)71A>G	None
New	Intron 10	(-)50A>G	None
rs17170590	Intron 10	(-)25G>A	None
rs17213431	10	1857G>A	None
rs79624523	Intron 13	(-24)T>C	None
rs138189874	13	2193G>A	None
New	14	2469G>C	Q823H
rs2302527	Intron 15	(-91)A>G	None
New	Intron 17	(-)4G>A	None
rs6957214	Intron 19	(-)54T>C	None
rs78643563	19 (Splice site intronic)	(-)3T>A	None
rs2052194	19 (Essential splice site)	(-)1G>A	None
1000 genomes	Intron 23	(-)7A>G	None
rs2052197	24 (3 primer UTR)	(+)3C>T	None
rs115284398	24 (3 primer UTR)	(+)4G>A	None

Supplementary Table 3: ANLN exon primers

Primer Name	Primer Sequence
ANLN-1F	CACTTTTCTCTTCCTGAATTTGAAC
ANLN-1R	TGACAGAGGAAGGTGGGTG
ANLN-2F	AAATTTGTGGCCGTAAAAATC
ANLN-2R	AATGAAATGTTTGGGGCTTG
ANLN-3F	TTTAAAAGAATAGGGAGGGGTG
ANLN-3R	ATGCAAGCAAAGGATACTCAAC
ANLN-4F	ATTCAGCATAGAGTGATCCTGGT
ANLN-4R	CCATCCACCTGCACATACAC
ANLN-5F	GGACTTGAATTGTTTTGTTATAGGAC
ANLN-5R	CAAATCATTGCTGTACCATTCA
ANLN-6F	CAAAGCATTTTGAAGCTGTAATG
ANLN-6R	GGCATCAGAACCCATTTTG
ANLN-7F	TCAGACAAGATTGGGCACAT
ANLN-7R	CGAAAAGTGACAGAGTTAATTGGA
ANLN-8F	CACTATCTCTTTGGTTCTAAGGAAAC
ANLN-8R	AGAACAAACAAATCCAGCAAAG
ANLN-9F	AAGAGAGGACAGGTGTTCAGG
ANLN-9R	CCCTGTCAAAGTCAGTGAGG
ANLN-10F	TTGAAGCTGAAGATTTTCTTGG
ANLN-10R	AGGTCTGCAAATTCCTTG
ANLN-11F	GGAGAATTCATTGATTTTCACAGA
ANLN-11R	TGTCAATCTAAACCATGACCCTTA
ANLN-12F	GGATAGTGCTCAGTGTGTTGC
ANLN-12R	AGCTCACAGCCTAGTGCAAG
ANLN-13F	TTTTGGTGCATAGTCGAGAAAC
ANLN-13R	TCCACTGGAACAGATGACTAGG
ANLN-14F	TTTGCTCTCATTAGAAACAGTTACG
ANLN-14R	ACAATTCAATCTAGGTGAGGTTCA
ANLN-15F	TTTGTGTCTGGAAAGTTGATTTTAG
ANLN-15R	GTGCATAAGGCGTTTCAAAG
ANLN-16.17F	AAATATTTTGGACTTGCATTATAGGG
ANLN-16.17R	AAATTGGAACATGAAACTGATCC
ANLN-18.19F	GGTTGGATAGTTTTACTTTCTGAGAC
ANLN-18.19R	TGCAAGTGCTTAATTCCTTACC
ANLN-20F	TTCTACTGGGATGGGGTGAG
ANLN-20R	AAAAGCATTGTGGCATTTC
ANLN-21F	TGCTCTGTTTTCAAGTTGTAATAGTC
ANLN-21R	AACAAGTCTGTATTTCAAAAATGG
ANLN-22F	CAGCATTTCATTGTTAGGACATTT
ANLN-22R	CAGAGGGAACATTTGCATGA
ANLN-23F	AAATGCTGCTTAATGCTTACTGAC
ANLN-23R	AGTGGTAAGTACATAGTGGGCAATC
ANLN-24F	TCCCTAGCAAGAGTACATGGG
ANLN-24R	TGCAATCAGTAAATCTGATGCTC

Supplementary Table 4: Anillin CDNA primers

Primer Name	Primer Sequence
VP1.5-F	GGACTTTCCAAAATGTCTG
ANLNcdna-1R	AGACACAGGACTTGGAGAAC
ANLNcdna-2F	ACGCTGTTCTGACAACACTGA
ANLNcdna-2R	TCCCTTTGGGAACAGAATGT
ANLNcdna-3F	ATTTGCTCCTGGGAAGATGA
ANLNcdna-3R	GGATGGCCTTTGTATTTGGA
ANLNcdna-4F	TCTGCAATCTCAATCTAAAGACAAA
ANLNcdna-4R	ACTGAGTTTTTTGAAACACCTTGG
ANLNcdna-5F	GAAAAAGGCGGAAACTCAA
ANLNcdna-5R	GTGCCAATGGTGCAAGTAA
ANLNcdna-6F	AGCCAAGAGGAGATGGATCA
ANLNcdna-6R	AAAGTGTTCTCTTCCCAGTTGC
ANLNcdna-7F	TCTATCAAGCTAGCCAGGCTCT
ANLNcdna-7R	GTGAGGAGTCGCTTTGGAGT
ANLNcdna-8F	ACAGCTTGGTGCAAAGAAAG
ANLNcdna-8R	TGCATTGGCTGACAAGAGTC
ANLNcdna-9F	CGCAAGAATCCCATAGGAAG
ANLNcdna-9R	GTTGTTGATGGCGTGCAG
ANLNcdna-10F	CTTCTACCACTTCGGCACCT
ANLNcdna-10R	CCTAGTCAGACAAAATGATGCAA