SUPPLEMENTAL DATA DESCRIPTION

The supplemental data contains 4 figures, 3 videos, 3 tables, and 1 file containing the figure/table/video legends.

Supplementary Figure Legends:

supplementary figure 1: Expression of *LAMA1* and other laminin subunits

(**A**) Quantitative real-time PCR results for *LAMA1* mRNA expression in multiple Human tissue cDNA panels (human MTC panels I and II and Human Fetal MT Panel; Clontech Laboratories, Mountain View, CA). Y axis represents the normalized relative expression of *LAMA1*.

(**B**) mRNA expression analysis of LAMB1, LAMB2, LAMC1 in Control, Patient 2, and Patient 3 fibroblasts by quantitative real-time PCR. Expression levels of *LAMB1*, *LAMB2*, *LAMC1* are lower in Patient 2 and Patient 3 as compared to Control. Error bars represent S.D. (n=5). *, $P<0.05$; **, $P<0.01$ (Mann-Whitney test to compare Patient 2 or Patient 3 to control).

supplementary figure 2. Optimization of *LAMA1* siRNA knockdown

To optimize the siRNA to be used in the study, we compared LAMA1 expression using different concentration of non-targeting and *LAMA1* siRNA. Since 0.6 μM showed the most efficient and consistent knockdown, it was used to assess evaluation of LAMA1 deficiency in cells.

supplementary figure 3. **Overexpression of full-length wild type LAMA1 rescues cell phenotype associated with LAMA1 deficiency.**

Rescue experiments were performed by stable transduction of wild type LAMA1 (pLenti-LAMA1) in control, Patient 2, and Patient 3 fibroblasts.

(**A**) Recovery of LAMA1 protein expression in Patient 2 and Patient 3 after transduction was done using immunoblotting. Vinculin (VCL) was used as loading control.

(**B**) Equal numbers of control and Patient 2 and 3 fibroblasts were plated on wells coated with either bovine serum albumin (BSA) or collagen-I (Col-I) or laminin-111 and allowed to attach (See also Figure 4A). Control and patients' fibroblast were transduced with a lentiviral vector containing the wild type form of *LAMA1* (pLenti_LAMA1) After 24 hours, cells were

trypsinized and transferred to non-coated wells and allowed to attach. OD values represent amounts of DNA in cells that remain attached to the wells after 90 minutes of incubation.

(C) Equal number of control Patient 2, and Patient 3 fibroblasts were seeded on culture wells and allowed to form a monolayer and grow to confluency, after which a 0.9 mm scratch was made. "Wound" closure was monitored by imaging cells stained with Calcein AM after 12, 24, and 36, 48, and 60 hours. Percent closure and migration rates were calculated from 6 experiments. Scale bar represents 100μm. "-" indicates that cells were transduced with pLenti-Empty construct, while "+" indicated transduction with pLenti_LAMA1 that contains full length *LAMA1*. * shows statistical significance between Patient 2 or Patient 3 and Control cells. (**D**) Apoptosis was quantified using Caspase-3/7 activity measurement at 30 hours after plating control (white bars), Patient 2 (light grey bars) cells, and Patient 3 (dark grey bars) with or without the addition of the known apoptosis inducer, Staurosporine. Results show an increased susceptibility of patients' cells to apoptosis at baseline and recovery after transduction with pLenti_LAMA1 that contains wild type *LAMA1*. "-" indicates that cells were transduced with pLenti-Empty construct, while "+" indicated transduction with pLenti_LAMA1 that contains wild –type *LAMA1*.

(**E**) GTPase activation assay of Cdc42 in control, Patient 2 and Patient 3 fibroblasts. Results show a significant recovery of the activated form (GTP bound) of Cdc42 in Patient 2 and Patient 3 fibroblasts after transduction with pLenti_LAMA1 containing wild type *LAMA1*.

For (**B**-**E**), error bars represent S.D. for experimental replicates (3 replicates for B, D, and E; 6 replicates for C). Analysis was done comparing patient cells transduced with pLenti-Empty and control, or patient cells transduced with pLenti-LAMA1 and control. * indicates $P<0.05$, Mann-Whitney test.

supplementary figure 4. LAMA1 immunoblot additional data.

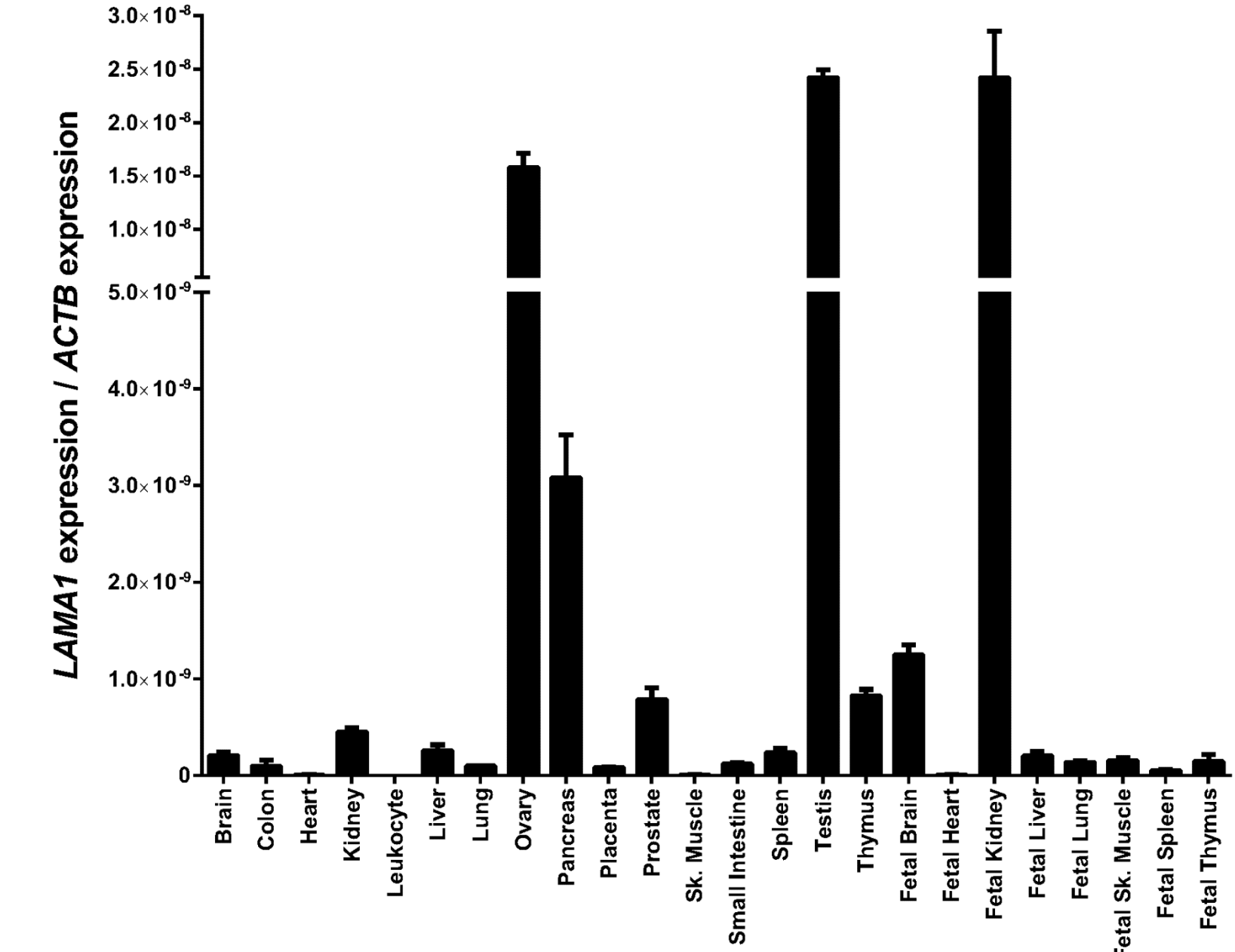
(**A**) Full immunoblot images for LAMA 1 and vinculin (shown in figure 2C). Corresponding molecular weight is shown in the ladder (HiMark Pre-stained Protein Standard).

(**B**) Full immunoblot images for LAMA 1 and vinculin (shown in supplementary figure 2A). Corresponding molecular weight is shown in the ladder (HiMark Pre-stained Protein Standard).

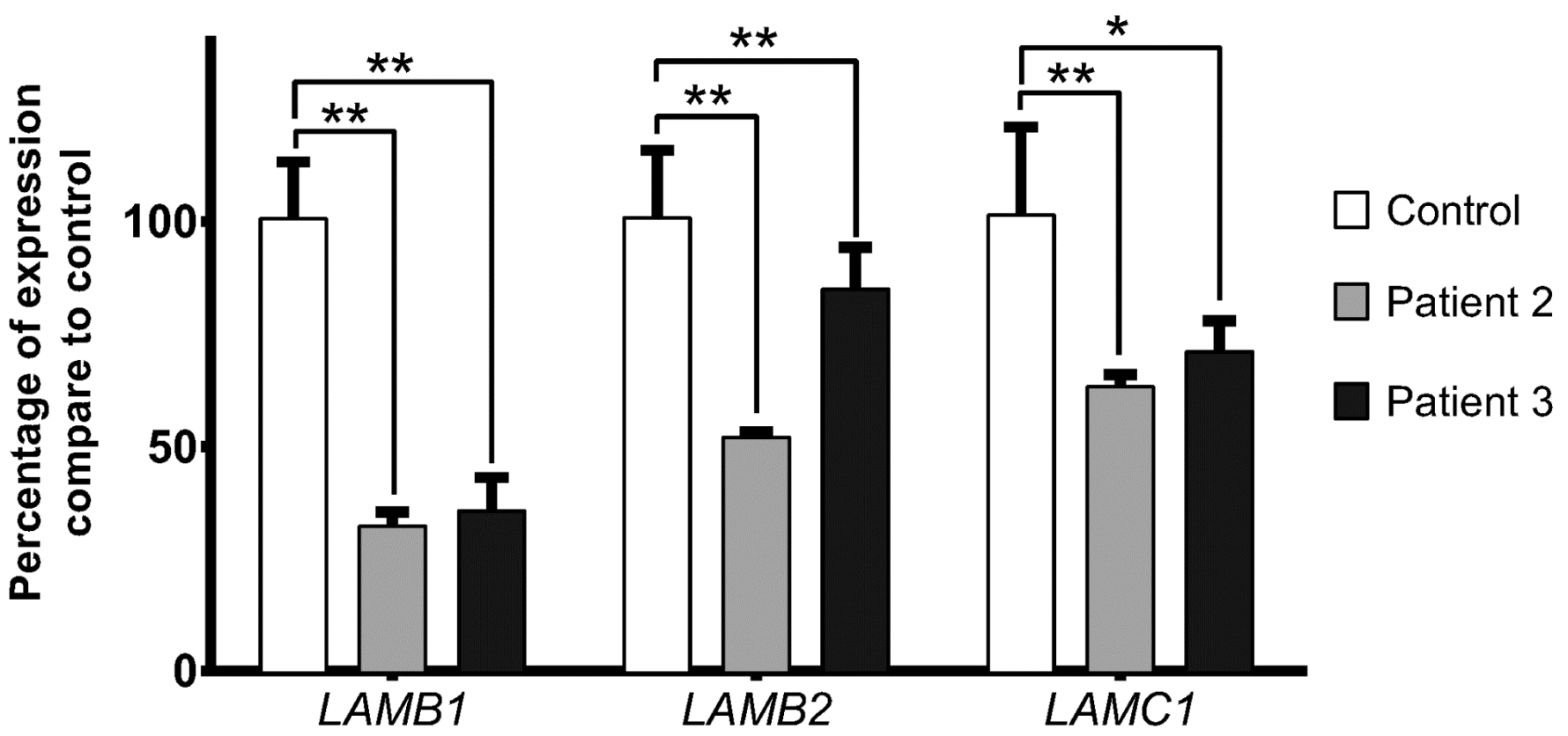
(**C**) LAMA1 antibody competition assay with laminin-111 protein, showing specificity of LAMA1 antibody used in this study. A total of 8 μg laminin-111 peptide was used.

Supplementary Figure 1

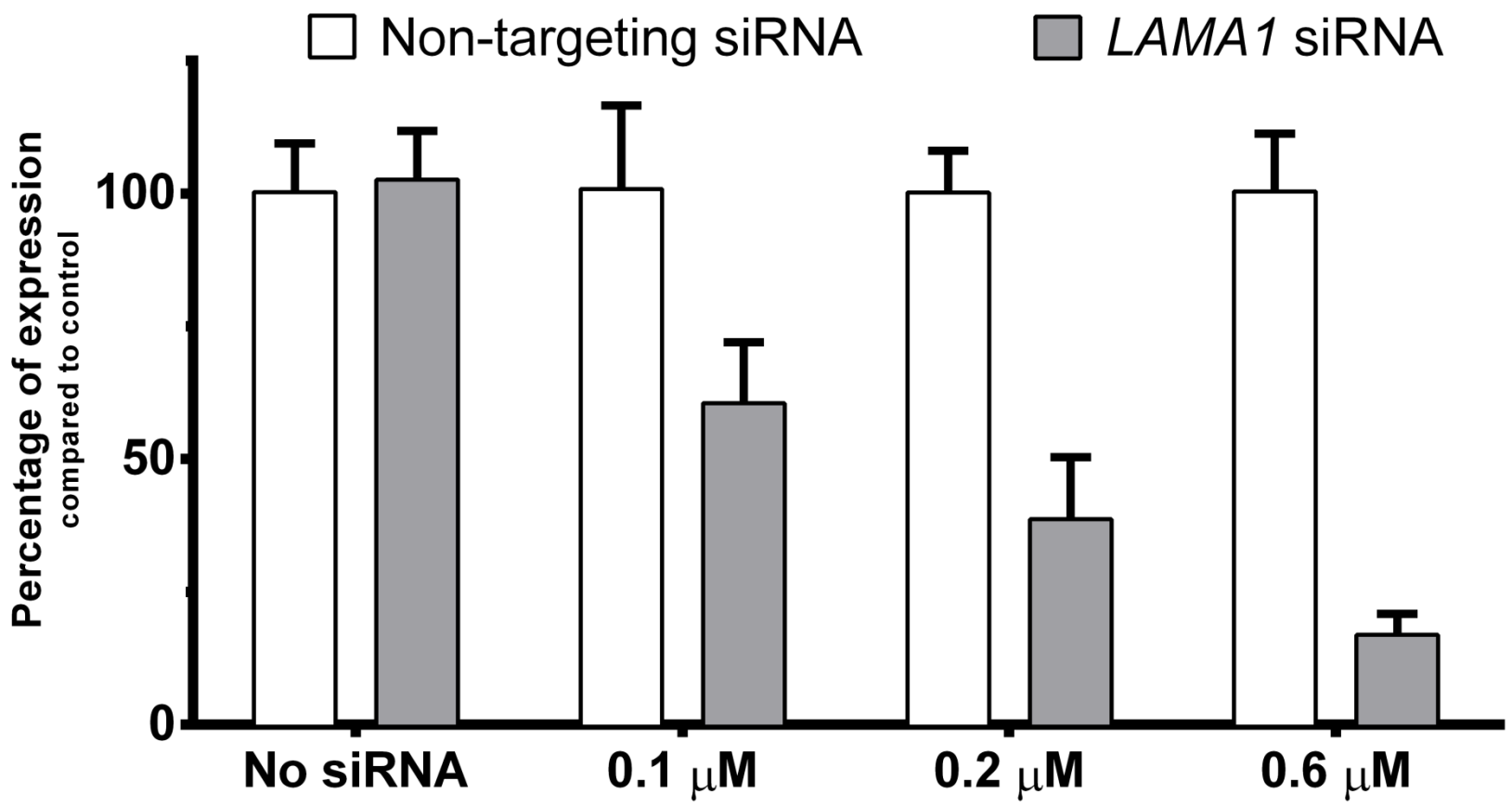
A



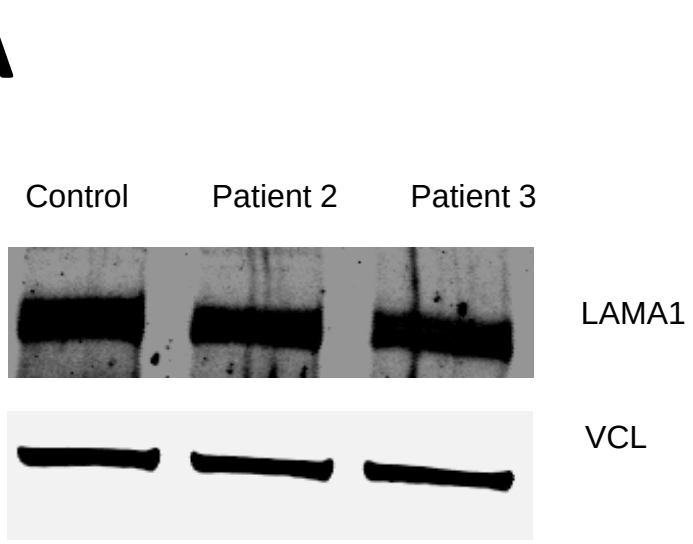
B

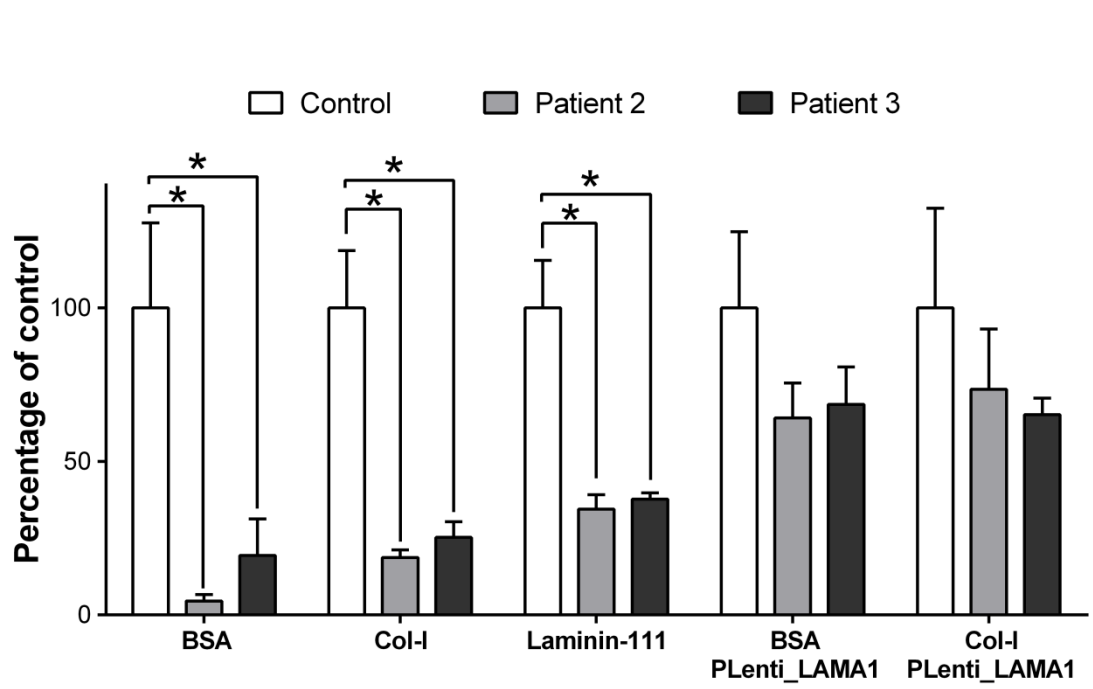


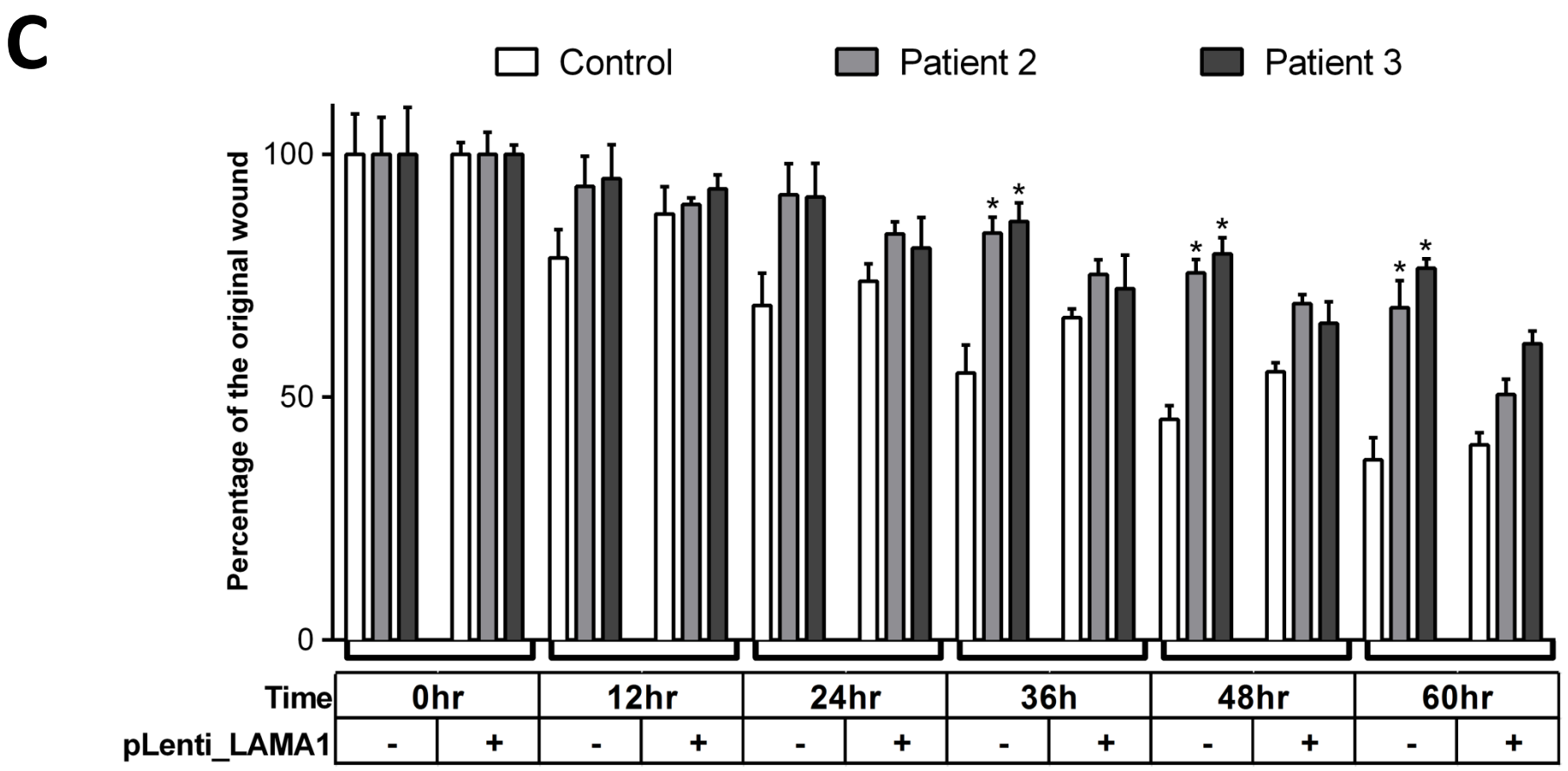
Supplementary Figure 2

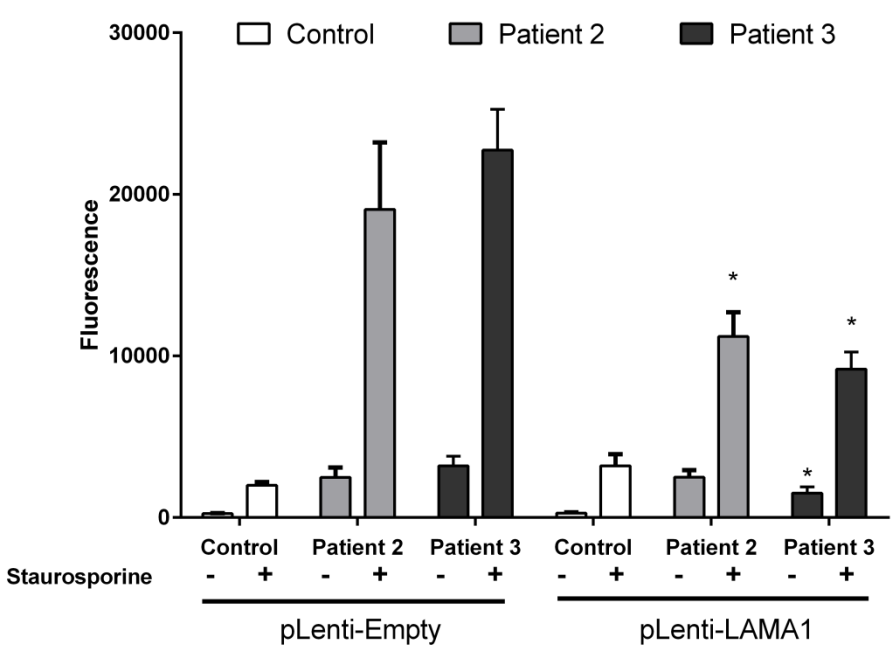


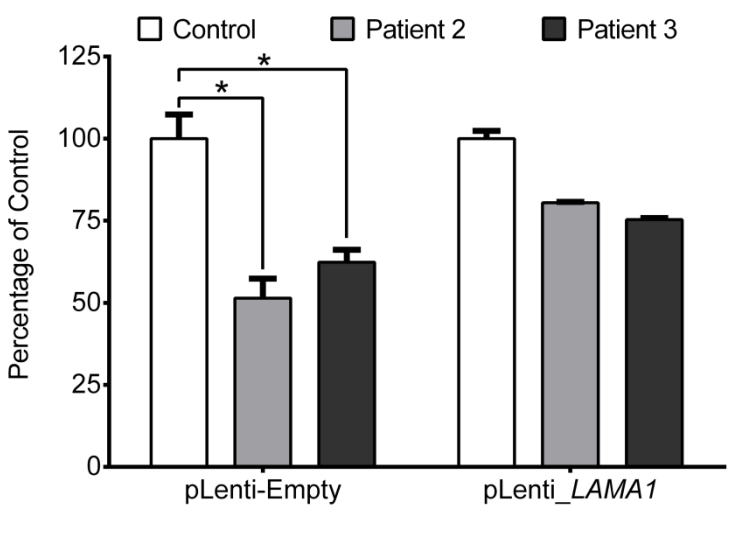
Supplementary Figure 3





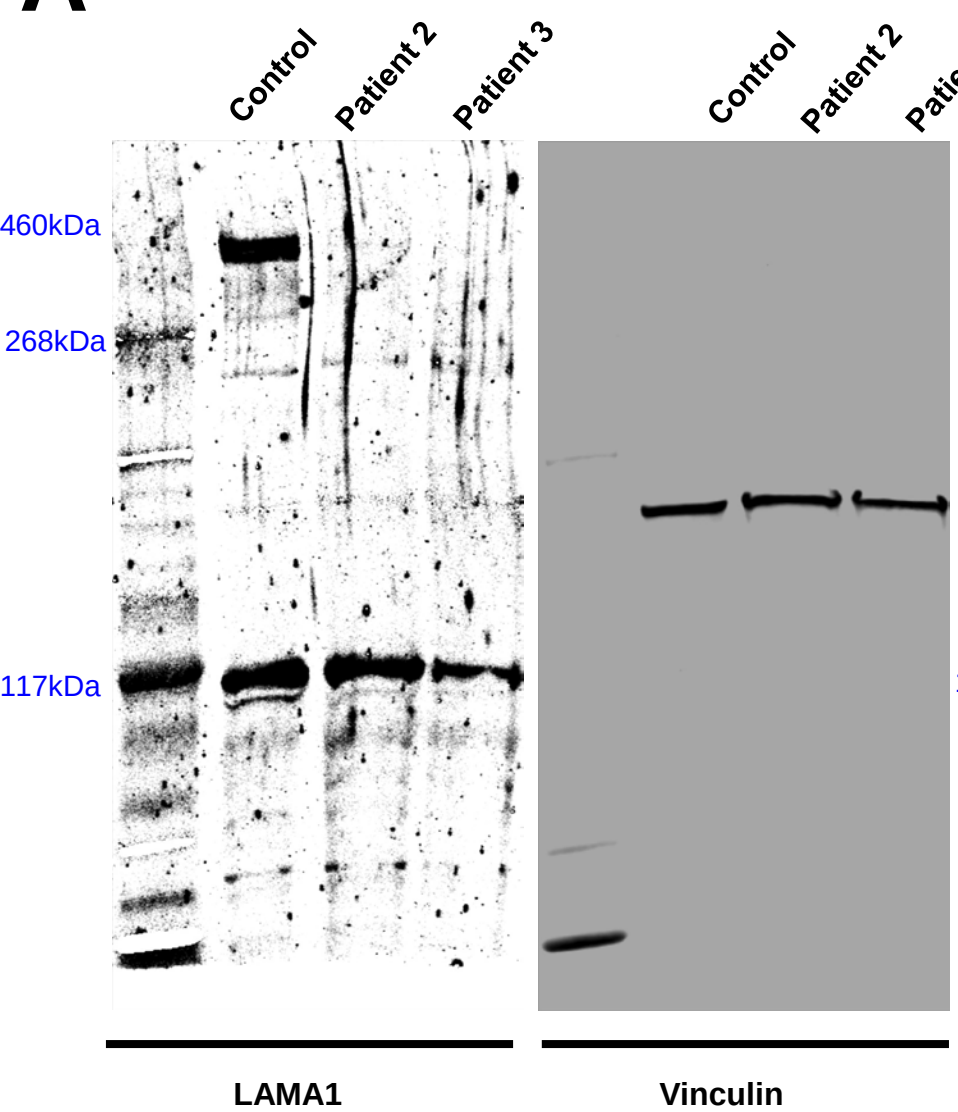






Supplementary Figure 4

A



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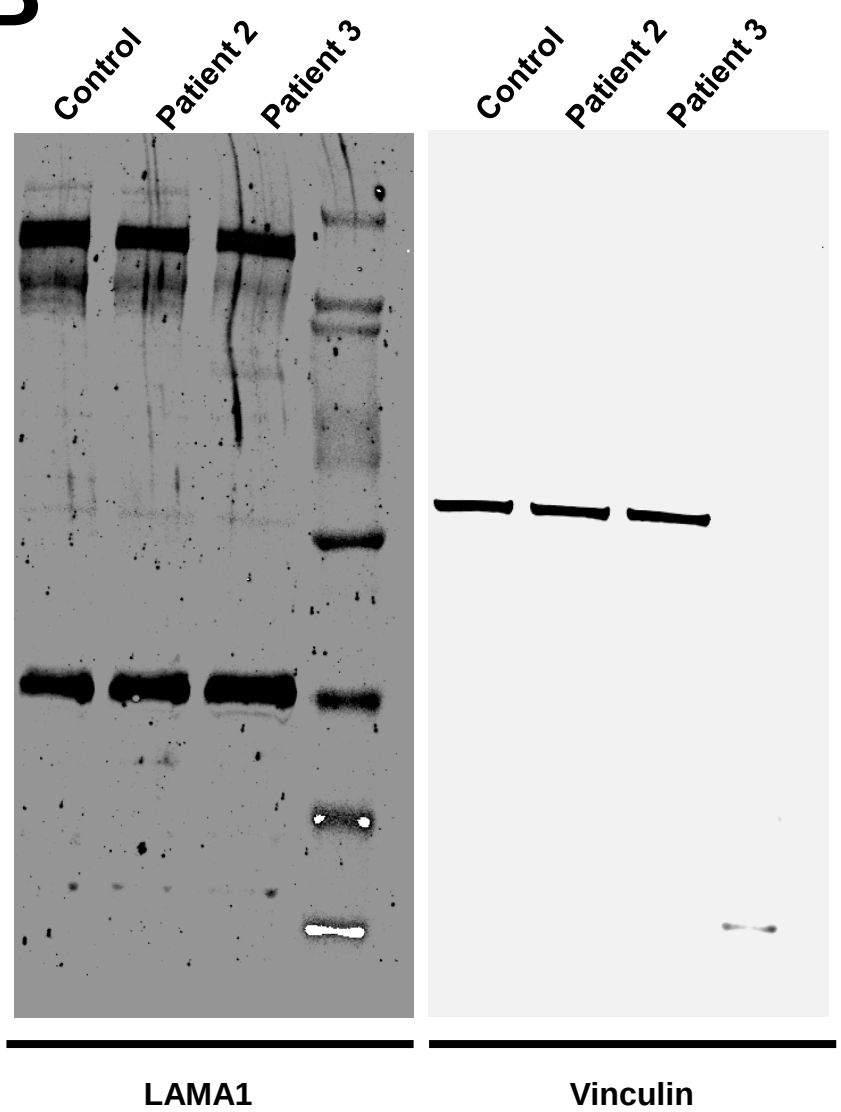

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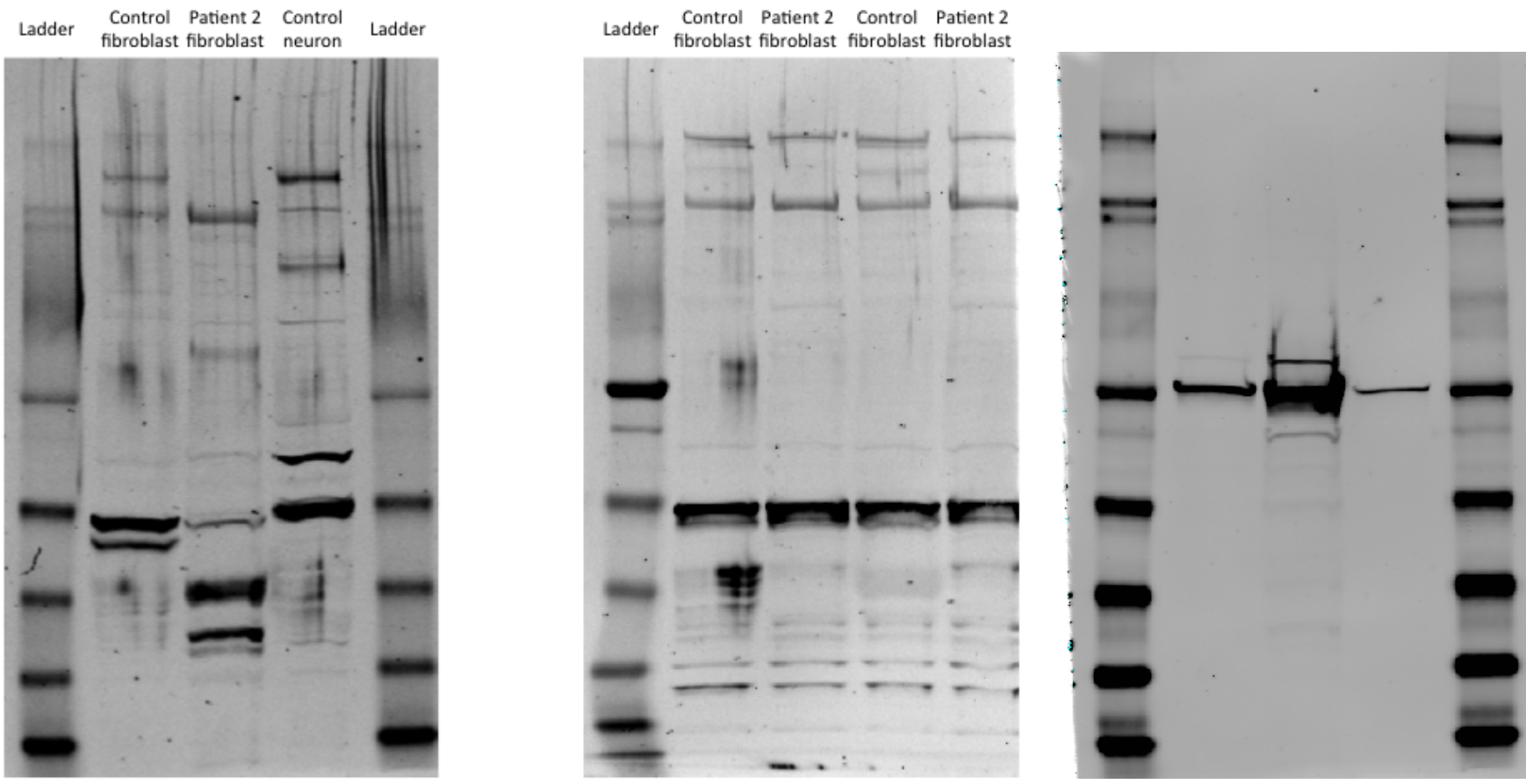

supplementary video 1:

Video illustration of involuntary tic movements in Patients 2 and Patient 3. In Patient 2, numerous simple tics are observed during casual examination. The movements are characterized by brief stereotyped "wrinkling" of the nose and elevation of the upper lip or eye rolling movements. Patient 3 movements are more complex, at times, in the form of complex bilateral "arm pumping" action. While both patients describe these movements as involuntary, both indicate a premonitory urge to move, which is relieved by the occurrence of the movements as typically seen in patients with motor tics.

supplementary Videos 2a (left eye) and 2b (right eye):

Video illustration of ocular motor assessment, gross abnormalities were observed on all tests with numerous saccadic intrusions documented during gaze holding.

Supplementary table 1. Human disorders caused by laminin defects.

Gene	Protein	Human Disease	Reference
LAMA1	Laminin α1	Cystic cerebellar dysplasia associated with degenerative myopia	[1], and present report
LAMA2	Laminin α2	Congenital muscular dystrophy with diffuse cerebral gray and white matter abnormalities	[2]
LAMA4	Laminin α4	Dilated cardiomyopathy	[3]
LAMB1	Laminin β1	Cobblestone lissencephaly without ocular or muscular abnormalities	[4]
LAMB2	Laminin β2	Nephrotic syndrome with or without ocular anomalies (Pierson syndrome)	[5]
LAMA3	Laminin α3	Epidermolysis bullosa	[6]
LAMB3	Laminin β3	Epidermolysis bullosa	[7]
LAMC1	Laminin γ1	Autosomal dominant Dandy-Walker malformation and occipital cephaloceles	[8]
LAMC2	Laminin γ2	Epidermolysis bullosa	[9]
LAMC3	Laminin γ3	Cerebral cortical malformations, occipital	[10]

Supplementary table 2: Genes associated with Tics and Anxiety

Disease	Genes and the defects associated Candidate genes associated (OMIM number)	Gene defects associated	Reference	Phenotype
Gilles De La Tourette Syndrome	*PVRL3* (607147)	Missense	[11]	Motor and vocal tics, Aggressive behavior
	MRPL3 (607118)	Missense		
	DNAJC13 (614334)	Missense		
	OFCC1 (614287)	Frameshift, rare 3'UTR variant		
	SLITRK1 (609678)	Gene disruption	[12]	
	IMMP2L (605977)	Gene disruption	[13]	
	CNTNAP2 (604569)	Missense	[14]	
	HDC (142704)	Hemizygous	[15]	
	NLGN4 (300427)	Deletion	[16]	
Primrose Syndrome	*ZBTB20* (606025)	Missense variations	[17]	Tics or anxieties, Aggression
Choreo-acanthocytosis	*VPS13A* (605978)	Several variants	[18], [19], [20]	Tics and anxieties, Dystonia
Obsessive-Compulsive Disorder	*BDNF* (113505	Several variants	[21]	Tic related Obsessive-compulsive disorder
	HTR2A (182135)	Promoter variant	[22]	
	SLC6A4 (182138)	Promoter variant Missense	[23], [24]	
Myoclonic Dystonia	*SGCE* (604149)	Several variants	[25]	Depression, Anxiety, Obsessive Compulsive Disorder
	DRD2 (126450)	Missense	[26], [27]	
	DYT1 (128100)	Small Deletion	[28]	
Basal Ganglia Calcification	*PDGFB* (190040)	Several variants	[29]	Motor tics, Dementia, Anxiety, Psychosis
Chorea, Benign Hereditary	*NKX2-1* (600635)	Whole gene deletion	[30]	Movements exacerbated by anxiety, Chorea
		Intron variant	[31]	

Supplementary table 3. Supplemental table 3. List of the potential null variants (HGVS nomenclature) identified in *LAMA1* (NM_005559.3) from different databases (1000Genomes: www.1000genomes.org, NHLBI Exome Sequencing Project: evs.gs.washington.edu and the Exome Aggregation Consortium: http://exac.broadinstitute.org) and their frequency when applicable. For each database, a frequency of loss of function (Corresponding to the sum of the frequencies) has been calculated. LoF: Loss of Function

Variant name in database	Frequency
Exome Variant Server (EVS)	
c.858+1G>T; p.?	0.000231
c.1492_1493insC; p.Arg498Profs*13	0.00304
c.2344C>T; p.Arg782*	0.000077
c.4601_4607del; p.Ala1534Glyfs*13	0.000479
c.5512C>T; p.Gln1838*	0.000077
c.6008-2A>G; p.?	0.000077
c.8188C>T; p.Gln2730*	0.000077
c.8737del; p.Asp2913Metfs*2	0.00008
LoF variants in EVS	**0.004138**
1000 Genomes (1KG)	
c.858+1G>T; p.?	0.000277
c.6476dup; p.Ser2160*	NA
c.7452+2T>G; p.?	NA
c.8207+2T>C; p.?	0.000458
c.8498_8499insA; p.Lys2832Glufs*4	NA
c.8501_8502insC; p.Leu2835Phefs*33	NA
c.8904_8905insCC; p.Ala2969Profs*24	0.000719
LoF variants in 1KG	**0.001454**

Exome Aggregation Consortium (ExAC)	
c.9084dup; p.Cys3029Metfs*19	0.000008291
c.9067+1G>A; p.?	0.00001655
c.8777del; p.Asn2926Metfs*17	0.000008239
c.8737del; p.Asp2913Metfs*2	0.000008241
c.8629dup; p.Val2877Glyfs*20	0.000008241
c.8608C>T; p.Gln2870*	0.000008243
c.8550_8556dup; p.Ile2853Trpfs*17	0.000008249
c.8556+1G>A: p.?	0.000008249
c.8208-1G>T; p.?	0.000008848
c.8207+2T>C; p.?	0.00001052
c.8192C>A; p.Ser2731*	0.000008837
c.8095-2dup; p.?	0.000008825
c.8094+1G>A; p.?	0.0000083
c.7900dup; p.Thr2634Asnfs*33	0.000008238
c.7779-1G>A; p.?	0.00001666
c.7779-2A>G; p.?	0.000008341
c.7627-1G>T; p.?	0.000008364
c.7514C>G; p.Ser2505*	0.000008271
c.7338-2dupA; p.?	0.000008238
c.7338-1G>T; p.?	0.000008238
c.7246C>T; p.Gln2416*	0.000008238
c.7243A>T; p.Lys2415*	0.000008239
c.7195+2T>A; p.?	0.000008237
c.7180C>T; p.Arg2394*	0.00002471

c.6899+2T>C; p.?	0.000008239
c.6517C>T; p.Arg2173*	0.000008455
c.6490-2A>G; p.?	0.000008985
c.6489+1del; p.?	0.000008241
c.6299C>G; p.Ser2100*	0.000008252
c.6190+2T>C; p.?	0.000008236
c.6008-2A>G; p.?	0.000008273
c.5796+1G>A; p.?	0.00001648
c.5706dup; p.Ala1903Serfs*12	0.000008238
c.5661-1G>A; p.?	0.000008249
c.5661-1G>T; p.?	0.000008249
c.5497-1G>A; p.?	0.000008238
c.5169-2A>G; p.?	0.000008299
c.5119C>T; p.Gln1707*	0.000008237
c.4957del; p.Glu1653Argfs*7	0.000008238
c.4897-1G>A; p.?	0.00001649
c.4896+2T>C; p.?	0.000008243
c.4579C>T; p.Gln1527*	0.00001659
c.4383-1G>C; p.?	0.000008713
c.4383-2A>C; p.?	0.000008768
c.4300_4303dup; p.Thr1435Metfs*15	0.00001655
c.4257>A; p.Cys1419*	0.0000171
c.4171_4172del; p.Arg1391Glyfs*19	0.0000251
c.4023_4032dup; p.Lys1345Glyfs*6	0.000008238
c.3919C>T; p.Arg1307*	0.00002472

c.3896_3897del; p.Ser1299Cysfs*3	0.000008255
c.3687+1G>T; p.?	0.00001116
c.3479C>G; p.Ser1160*	0.0000131
c.3476del; p.Cys1159Serfs*8	0.00001295
c.3450C>A; p.Cys1150*	0.00001092
c.3397C>T; p.Arg1133*	0.00001765
c.3364-1G>A; p.?	0.00001729
c.3099G>A; p.Trp1033*	0.000008238
c.3053dup; p.His1019Serfs*8	0.000008237
c.3021C>A; p.Cys1007*	0.000008237
c.2986del; p.Thr996Hisfs*28	0.00002472
c.2935del; p.Arg979Glyfs*45	0.00000824
c.2791C>T; p.Gln931*	0.000008604
c.2754del; p.Leu920Serfs*8	0.000008336
c.2702-1G>T; p.?	0.000008993
c.2487del; p.Arg830Aspfs*42	0.000008509
c.2403-2A>G; p.?	0.000009118
c.2344C>T; p.Arg782*	0.00003628
c.1957C>T; p.Gln653*	0.000008237
c.1885C>T; p.Gln629*	0.000008237
c.1583G>A; p.Trp528*	0.000008249
c.976+2T>C; p.?	0.000008237
c.891C>A; p.Cys297*	0.000008238
c.858+1G>T; p.?	0.00009914
c.733G>T; p.Glu245*	0.000008238

c.730del; p.Arg244Glyfs*18	0.000008238
c.505C>T; p.Arg169*	0.00001648
c.448del; p.Gln150Serfs*16	0.000008238
c.425del; p.Asp142Valfs*24	0.00000824
c.404G>A; p.Trp135*	0.000008247
c.391C>T; p.Arg131*	0.000008264
c.381_382del; p.Asn128Cysfs*49	0.000008278
c.307G>T; p.Glu103*	0.000008256
c.184C>T; p.Arg62*	0.000008247
c.11dup; p.Val5Argfs*66	0.00001018
LoF variants in ExAC	**0.000988909**

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