SUPPLEMENTARY MATERIAL

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Supplementary Methods

Fibroblast culture and induced SRRM4 expression

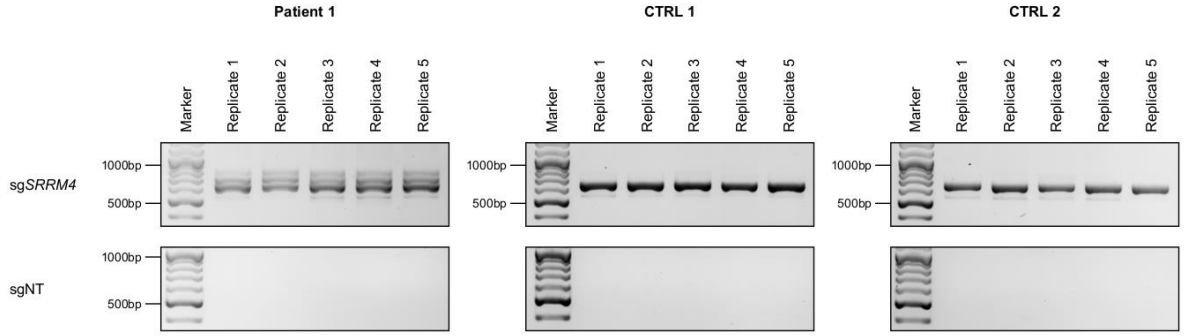
Fibroblast lines established from skin-biopsy specimens were cultured in DMEM (Gibco #41966052) supplemented with 10% FBS, Penicillin-Streptomycin (Gibco #15140122) and 0.2 mM uridine at 37°C, 5% CO_2 in a humidified atmosphere incubator, as described[2]. To activate *SRRM4* expression in fibroblasts, we used a lentiviral CRISPR-activation (CRISPRa) system[3], expressing a specific guide RNA (sg*SRRM4*, 5'-GAGGGATCAGCGCTGCAACT-3') directed to the promoter region of the canonical isoform of *SRRM4* (NM_194286.4). A non-targeting guide (sgNT, 5'-ACGGAGGCTAAGCGTCGCAA-3') was used as control. The CRISPRa system was comprised of three transfer vectors, namely lenti sgRNA(MS2)_puro optimized backbone, lenti dCAS-VP64_Blast, and lentiMPHv2 (Addgene plasmids # 73797, # 61425 and # 89308, gifts from Feng Zhang). Lentivirus was generated as previously described[4], using envelope plasmids pCAG_Eco (Addgene Plasmid #35617, gift from Arthur Nienhuis & Patrick Salmo) or pMD2.G, together with psPAX2 packaging plasmid (Addgene Plasmids #12259 and #12260, gifts from Didier Trono). For sgRNA(MS2)_puro optimized backbone we generated ecotropic murine lentivirus using pCAG_Eco. Fibroblasts were seeded at 5,000 cells per well in a 24 well plate 24h prior to transduction. Cells were then co-transduced with three lentiviruses using the 5 µl of Ecotropic Receptor Booster (Takara # 631471) per well, and treated according to the manufacturer's protocol. 48h after infection, 75 µg/ml hygromycin, 5 µg/ml blasticidine and 1 µg/ml puromycin were added to the culture media for 7 days, to select for triple-infected cells. Cells were passaged 3 times, expanded until 90% confluency on a 10 cm dish was reached and harvested for RNA extraction.

Verification of SRRM4 expression

Isolated RNA was reverse-transcribed using the High-Capacity cDNA Reverse Transcription Kit (Applied Biosystems). Expression of *SRRM4* was then assessed by PCR using primers spanning exons 2-7 (*SRRM4*-F: 5'-CAGAGCCCCAGAATAACCCC-3', *SRRM4*-R: 5'-GCTGACAGGTAGCCAAGCAT-3'), followed by agarose gel electrophoresis

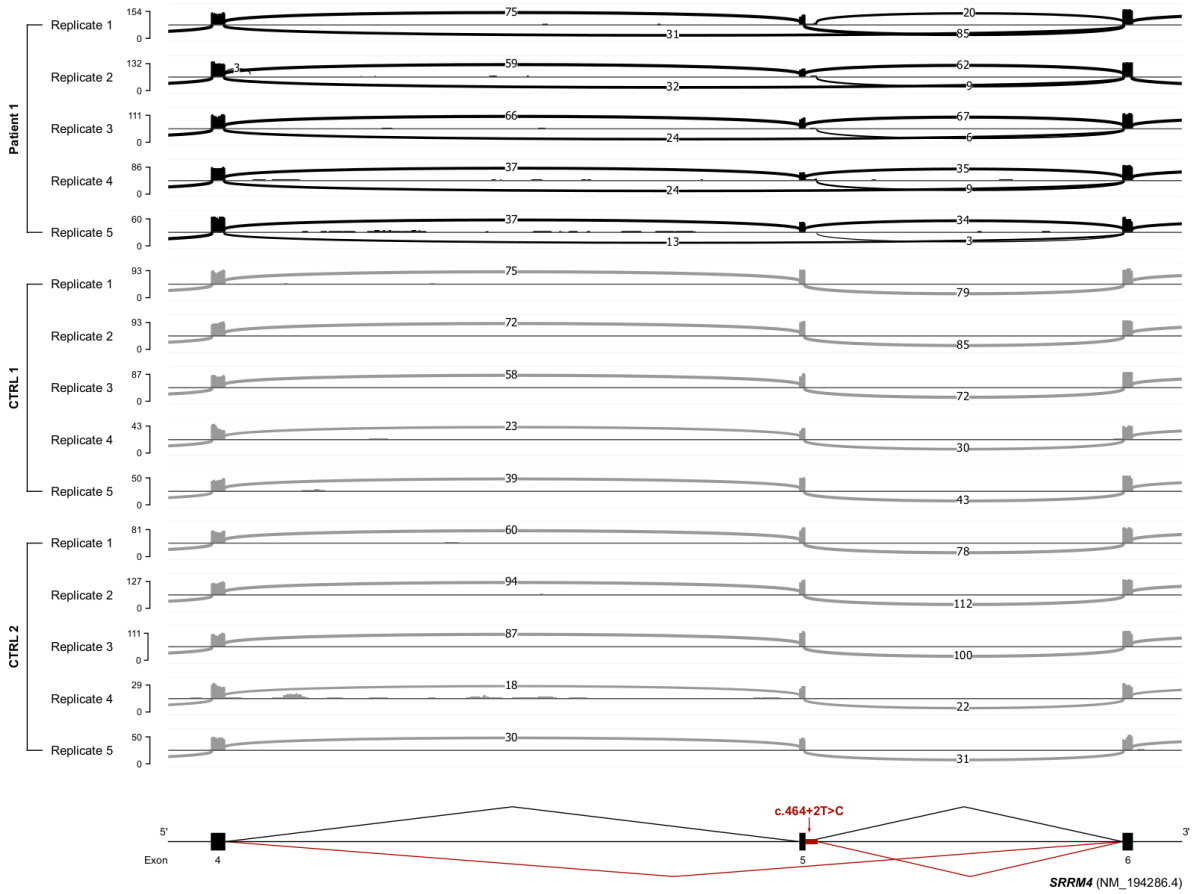
Supplementary Figures

Suppl. Figure 1 cDNA analysis of *SRRM4* expression following CRISPR activation (CRISPRa)



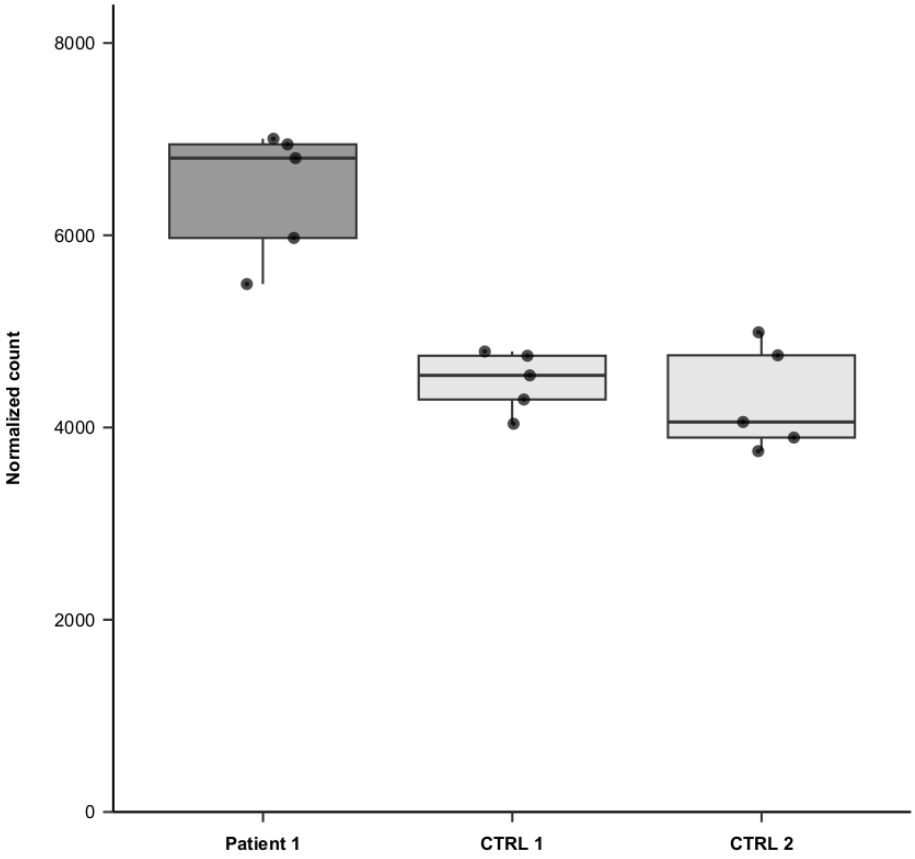
Agarose gels showing *SRRM4* RT-PCR amplicons produced from cDNA of *SRRM4*-activated fibroblast cell lines (sg*SRRM4*) from patient 1 and the control subjects (CTRL 1, CTRL 2). Five biological replicates were analyzed for each subject (patient 1, CTRL 1, CTRL 2). RT-PCT primers were designed to span *SRRM4* exons 2-7. Note the presence of three different RT-PCR products for patient 1. No RT-PCR amplicons were detected in non-*SRRM4*-activated fibroblast cell lines (sgNT) from the three subjects.

Suppl. Figure 2 *SRRM4* pre-mRNA splicing consequences associated with patient 1`s recurrent c.464+2T>C variant across all biological replicates



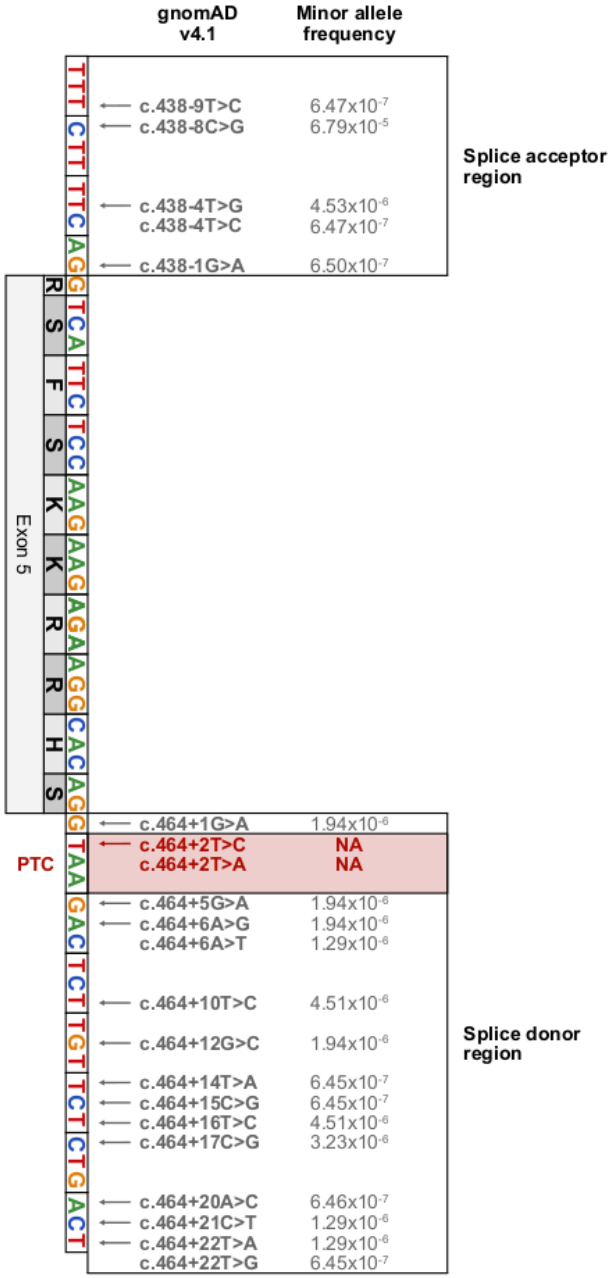
Sashimi plots established from srRNA-seq data generated for *SRRM4*-activated fibroblast cell lines from patient 1 and the control subjects (CTRL 1, CTRL 2) are shown. Five biological replicates were analyzed for each subject (patient 1, CTRL 1, CTRL 2). CRISPRa, CRISPR activation; srRNA-seq, short-read RNA sequencing.

Suppl. Figure 3 Short-read RNA sequencing (srRNA-seq) data-based quantification of *SRRM4* expression following CRISPR activation (CRISPRa)



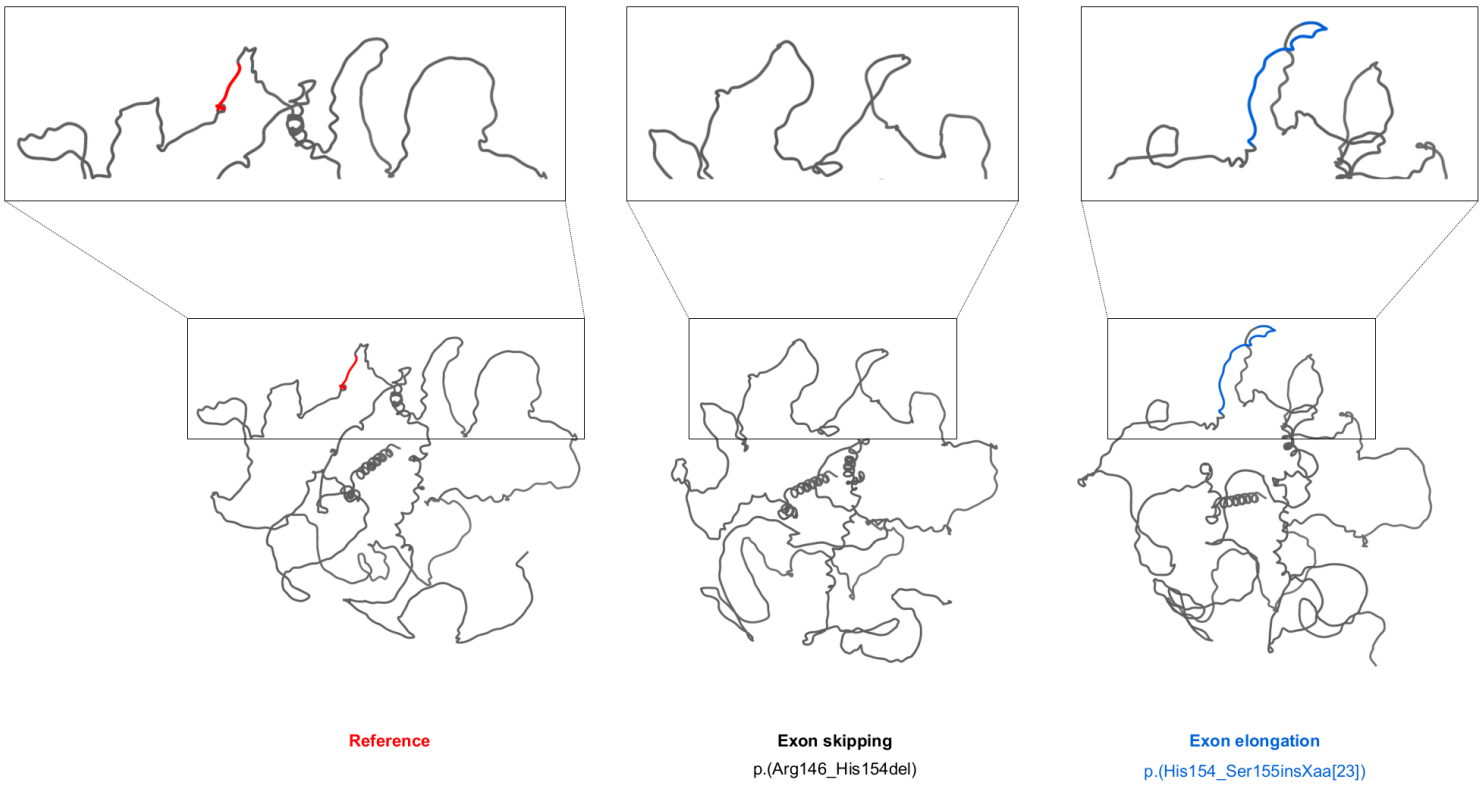
Box plots showing the results from quantitative analysis of *SRRM4* expression in *SRRM4*-activated fibroblast cell lines from patient 1 and the control subjects (CTRL 1, CTRL 2). For the quantifications, srRNA-seq data were analyzed using DESeq2[5]. Five biological replicates were included in the analysis for each subject (patient 1, CTRL 1, CTRL 2).

Suppl. Figure 4 A premature termination codon (PTC)-encoding TAA triplet in *SRRM4* intron 5 is not affected by variants in gnomAD v4.1



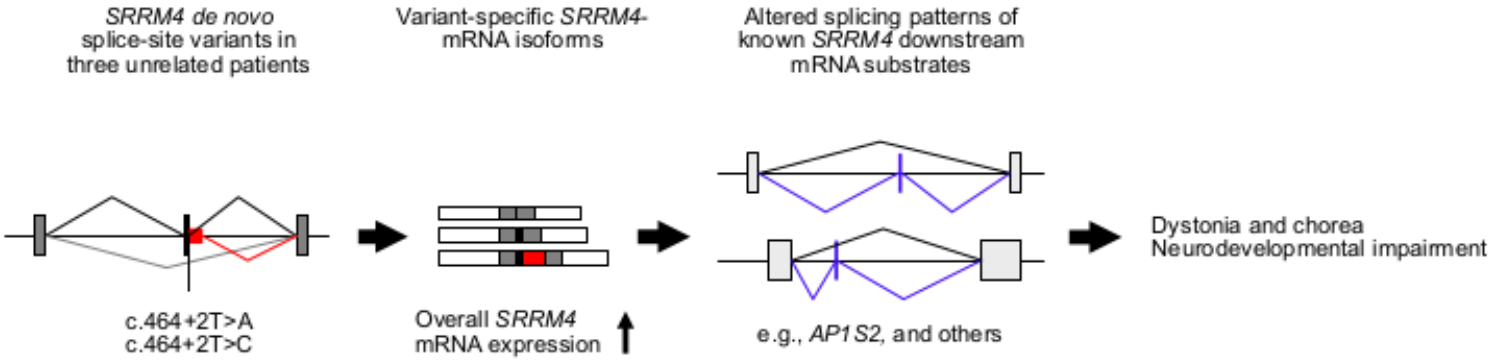
The patient-specific c.464+2T>C and c.464+2T>A variants were predicted to affect the invariant PTC. Note gnomAD v4.1 variants at positions +1, +5, and +6 but not at the PTC-encoding positions +2, +3, and +4. NA, not available.

Suppl. Figure 5 Predicted protein consequences associated with patient 1`s recurrent *SRRM4* c.464+2T>C variant



Structural effects on a 3D model of SRRM4 predicted by AlphaFold2[6] and visualized by UCSF ChimeraX[7] are illustrated for the predicted protein changes resulting from the mutation of the splice-donor site of intron 5 of *SRRM4*. The affected protein regions are highlighted in the zoom-in panels, with annotations of the predicted protein effects as follows (annotations based on NP_919262.2): exon skipping, p.(Arg146_His154del); exon elongation, p.(His154_Ser155insXaa[23]).

Suppl. Figure 6 Schematic depiction of proposed *SRRM4* variant-associated disease pathogenesis



The altered *SRRM4* splice site is associated with new *SRRM4* mRNA-isoforms and increased overall *SRRM4* mRNA levels, consistent with a potenital toxic gain-of-function effect, and resulting downstream perturbation of alternative splicing control.

Supplementary Tables

Suppl. Table 1 List of 131 significant splicing-in events (FDR<0.05 and ΔPSI >0.05) in 111 genes in *SRRM4*-activated control fibroblasts compared to their non-*SRRM4*-activated counterparts (rMATS-turbo analysis[1])

Gene ID	Gene symbol	Chromosome	Exon start	Exon end	P value	FDR	Inclusion level difference	Raj et al.[8] N2A [gene]	Raj et al.[8] N2A [event]
ENSG00000116337.15_5	*AMPD2*	chr1	110167924	110168409	6.63E-05	0.016444242	0.26	No	No
ENSG00000118200.14_3	*CAMSAP2*	chr1	200797700	200797733	4.14E-06	0.001671941	0.088	No	No
ENSG00000116406.18_3	*EDEM3*	chr1	184670630	184670678	8.32E-06	0.002950034	0.111	No	No
ENSG00000116406.18_3	*EDEM3*	chr1	184670663	184670678	2.05E-04	0.039612673	0.088	No	No
ENSG00000155816.19_3	*FMN2*	chr1	240343468	240343480	5.11E-15	3.00E-11	0.134	No	No
ENSG00000174842.16_2	*GLMN*	chr1	92732256	92732298	1.93E-04	0.038061979	0.157	No	No
ENSG00000054523.17_4	*KIF1B*	chr1	10333071	10333089	1.56E-05	0.004967893	0.12	Yes	Yes
ENSG00000163875.15_3	*MEAF6*	chr1	37962307	37962337	3.46E-07	2.24E-04	0.215	Yes	Yes
ENSG00000116604.17_4	*MEF2D*	chr1	156446285	156446306	1.01E-11	3.07E-08	0.192	Yes	Yes
ENSG00000097033.14_3	*SH3GLB1*	chr1	87194085	87194172	2.12E-11	5.89E-08	0.053	Yes	Partial
ENSG00000097033.14_3	*SH3GLB1*	chr1	87194085	87194124	7.88E-10	1.37E-06	0.054	Yes	Yes
ENSG00000097033.14_3	*SH3GLB1*	chr1	87195770	87195794	1.11E-16	8.70E-13	0.083	Yes	Yes
ENSG00000115657.13_4	*ABCB6*	chr2	220082391	220082529	7.49E-05	0.018190743	0.225	No	No
ENSG00000143933.16_3	*CALM2*	chr2	47399552	47399598	2.20E-05	0.006669344	0.192	No	No
ENSG00000172292.14_3	*CERS6*	chr2	169622831	169622855	1.15E-06	5.99E-04	0.144	No	No
ENSG00000204843.12_3	*DCTN1*	chr2	74590740	74590755	0	0	0.159	Yes	Yes
ENSG00000144036.15_3	*EXOC6B*	chr2	72410022	72410034	7.22E-05	0.017668463	0.102	No	No
ENSG00000115414.18_4	*FN1*	chr2	216236631	216237045	1.50E-05	0.004791491	0.237	No	No
ENSG00000115419.12_4	*GLS*	chr2	191777918	191778090	5.18E-05	0.013524972	0.141	No	No
ENSG00000144366.15_6	*GULP1*	chr2	189454466	189454477	2.79E-04	0.047487563	0.183	No	No
ENSG00000151690.14_3	*MFSD6*	chr2	191364557	191364582	7.57E-06	0.002757969	0.154	Yes	Yes
ENSG00000061676.14_3	*NCKAP1*	chr2	183889705	183889723	0	0	0.248	No	No
ENSG00000189223.14_3	*PAX8-AS1*	chr2	114012966	114013126	2.98E-06	0.001303145	0.338	No	No

ENSG00000189223.14_3	*PAX8-AS1*	chr2	114016840	114017161	7.74E-06	0.002797177	0.222	No	No
ENSG00000055917.15_4	*PUM2*	chr2	20457997	20458131	3.79E-06	0.001569435	0.416	No	No
ENSG00000115694.14_3	*STK25*	chr2	242447420	242447550	1.09E-04	0.024363291	0.248	No	No
ENSG00000060971.17_3	*ACAA1*	chr3	38167652	38168115	8.21E-05	0.019641849	0.059	No	No
ENSG00000060971.17_3	*ACAA1*	chr3	38168000	38168115	9.79E-07	5.38E-04	0.373	No	No
ENSG00000060971.17_3	*ACAA1*	chr3	38168000	38168186	1.12E-06	5.93E-04	0.373	No	No
ENSG00000060971.17_3	*ACAA1*	chr3	38168000	38168200	1.12E-06	5.93E-04	0.373	No	No
ENSG00000114331.13_4	*ACAP2*	chr3	195046172	195046184	1.07E-05	0.003699884	0.12	Yes	Yes
ENSG00000161203.13_3	*AP2M1*	chr3	183898432	183898438	2.01E-04	0.039200009	0.102	No	No
ENSG00000163539.16_3	*CLASP2*	chr3	33615964	33615988	2.22E-05	0.006706592	0.16	Yes	Yes
ENSG00000114735.9_3	*HEMK1*	chr3	50609507	50609639	2.44E-04	0.044080821	0.167	No	No
ENSG00000114670.13_2	*NEK11*	chr3	130887673	130887781	1.12E-05	0.00379335	0.225	No	No
ENSG00000174748.20_5	*RPL15*	chr3	23959023	23959340	8.01E-05	0.019201791	0.151	No	No
ENSG00000174748.20_5	*RPL15*	chr3	23959327	23959340	1.70E-04	0.034569245	0.226	No	No
ENSG00000163697.16_4	*APBB2*	chr4	40859042	40859048	4.89E-05	0.012966296	0.123	No	No
ENSG00000181982.18_2	*CCDC149*	chr4	24822610	24822709	4.56E-05	0.012282297	0.08	No	No
ENSG00000181982.18_2	*CCDC149*	chr4	24822676	24822709	1.00E-08	1.05E-05	0.171	No	No
ENSG00000075539.14_4	*FRYL*	chr4	48504844	48504862	8.22E-06	0.002936096	0.164	Yes	Yes
ENSG00000084093.16_2	*REST*	chr4	57793780	57793830	3.51E-06	0.001487736	0.141	No	No
ENSG00000151612.15_4	*ZNF827*	chr4	146684241	146684274	5.80E-07	3.44E-04	0.517	No	No
ENSG00000113108.19_3	*APBB3*	chr5	139941428	139941434	1.37E-07	1.02E-04	0.402	No	No
ENSG00000081189.15_5	*MEF2C*	chr5	88026027	88026051	4.87E-06	0.001915515	0.227	No	No
ENSG00000120708.16_3	*TGFBI*	chr5	135389631	135390105	1.72E-04	0.034697329	0.093	No	No
ENSG00000047932.13_4	*GOPC*	chr6	117898610	117898634	5.18E-10	9.95E-07	0.254	Yes	Yes
ENSG00000135318.11_3	*NT5E*	chr6	86201694	86201816	9.03E-06	0.003179608	0.358	No	No
ENSG00000152894.14_5	*PTPRK*	chr6	128322340	128322352	2.88E-07	1.91E-04	0.104	No	No
ENSG00000152894.14_5	*PTPRK*	chr6	128324341	128324377	4.84E-06	0.001910863	0.09	No	No
ENSG00000198818.9_2	*SFT2D1*	chr6	166744872	166744895	1.09E-07	8.26E-05	0.131	No	No
ENSG00000111850.10_3	*SMIM8*	chr6	88040409	88040430	2.42E-04	0.044080821	0.187	No	No
ENSG00000285953.1_1	*AC000120.3*	chr7	91874215	91874448	2.15E-04	0.040538607	0.483	No	No

ENSG00000075624.14_4	*ACTB*	chr7	5567363	5567554	2.97E-04	0.049707554	0.544	No	No
ENSG00000133612.18_2	*AGAP3*	chr7	150835229	150835400	7.72E-05	0.018654616	0.192	No	No
ENSG00000153956.15_3	*CACNA2D1*	chr7	81612629	81612650	2.31E-07	1.60E-04	0.155	No	No
ENSG00000106554.12_4	*CHCHD3*	chr7	132571733	132571748	0	0	0.196	No	No
ENSG00000049540.16_4	*ELN*	chr7	73480273	73480327	1.36E-05	0.004407993	0.225	No	No
ENSG00000006652.13_4	*IFRD1*	chr7	112102324	112102433	6.39E-05	0.016070293	0.139	No	No
ENSG00000091127.13_2	*PUS7*	chr7	105137400	105137418	2.69E-05	0.007976862	0.134	Yes	Yes
ENSG00000127990.17_4	*SGCE*	chr7	94217089	94217124	0	0	0.1	Yes	Yes
ENSG00000153317.14_3	*ASAP1*	chr8	131173030	131173039	7.07E-13	3.16E-09	0.121	Yes	Yes
ENSG00000198363.17_3	*ASPH*	chr8	62594997	62595042	0	0	0.183	Yes	Yes
ENSG00000169398.19_4	*PTK2*	chr8	141772466	141772487	7.87E-13	3.36E-09	0.164	Yes	Yes
ENSG00000078668.13_3	*VDAC3*	chr8	42254195	42254198	3.01E-11	7.86E-08	0.135	No	No
ENSG00000165802.22_5	*NSMF*	chr9	140350080	140350086	8.80E-08	6.84E-05	0.109	No	No
ENSG00000119396.10_3	*RAB14*	chr9	123952831	123953008	1.75E-04	0.035128782	0.051	No	No
ENSG00000148341.17_3	*SH3GLB2*	chr9	131771731	131771746	4.03E-14	2.23E-10	0.113	Yes	Yes
ENSG00000197694.15_5	*SPTAN1*	chr9	131371929	131371944	0	0	0.161	Yes	Yes
ENSG00000197579.7_2	*TOPORS*	chr9	32550771	32550966	9.54E-05	0.021926665	0.236	No	No
ENSG00000160293.16_4	*VAV2*	chr9	136652367	136652382	8.82E-05	0.02087501	0.12	Yes	Yes
ENSG00000160293.16_4	*VAV2*	chr9	136675312	136675327	3.41E-05	0.009773681	0.105	Yes	Yes
ENSG00000107897.18_3	*ACBD5*	chr10	27512135	27512168	9.14E-05	0.021477637	0.191	Yes	Yes
ENSG00000107863.17_4	*ARHGAP21*	chr10	24911661	24911691	4.20E-11	1.07E-07	0.132	No	No
ENSG00000107863.17_4	*ARHGAP21*	chr10	24911661	24911804	4.30E-07	2.64E-04	0.056	No	No
ENSG00000148634.15_3	*HERC4*	chr10	69718869	69718893	2.01E-13	1.05E-09	0.138	Yes	Yes
ENSG00000096746.17_2	*HNRNPH3*	chr10	70098259	70098444	4.72E-05	0.012572793	0.066	No	No
ENSG00000197746.13_3	*PSAP*	chr10	73583644	73583650	5.55E-16	4.01E-12	0.125	Yes	Partial
ENSG00000197746.13_3	*PSAP*	chr10	73583644	73583653	2.19E-04	0.041019046	0.205	Yes	Yes
ENSG00000151532.13_2	*VTI1A*	chr10	114293288	114293309	3.75E-08	3.23E-05	0.209	Yes	Yes
ENSG00000166313.18_5	*APBB1*	chr11	6423206	6423212	1.58E-11	4.63E-08	0.092	Yes	Yes
ENSG00000167996.15_3	*FTH1*	chr11	61732869	61733001	2.13E-04	0.040477528	0.523	No	No
ENSG00000110514.19_3	*MADD*	chr11	47330530	47330593	1.27E-04	0.027592416	0.054	Yes	Yes

ENSG00000131626.18_5	*PPFIA1*	chr11	70197099	70197129	2.11E-05	0.006471834	0.501	Yes	Yes
ENSG00000131626.18_5	*PPFIA1*	chr11	70197099	70197129	3.19E-05	0.009273057	0.091	Yes	Yes
ENSG00000154134.14_2	*ROBO3*	chr11	124747861	124748332	2.71E-04	0.046438405	0.276	No	No
ENSG00000154134.14_2	*ROBO3*	chr11	124748193	124748332	2.45E-04	0.044080821	0.27	No	No
ENSG00000102189.16_2	*EEA1*	chr12	93240037	93240052	2.79E-04	0.047487563	0.066	Yes	Yes
ENSG00000139436.20_4	*GIT2*	chr12	110405118	110405124	1.09E-08	1.12E-05	0.159	Yes	Yes
ENSG00000051825.14_5	*MPHOSPH9*	chr12	123712009	123712163	5.67E-06	0.002175976	0.361	No	No
ENSG00000136021.18_3	*SCYL2*	chr12	100711458	100711470	4.90E-05	0.012966296	0.074	No	No
ENSG00000182796.14_4	*TMEM198B*	chr12	56225040	56225133	1.38E-04	0.029561537	0.244	No	No
ENSG00000180776.15_5	*ZDHHC20*	chr13	21967097	21967133	1.33E-06	6.74E-04	0.122	Yes	Yes
ENSG00000198513.11_3	*ATL1*	chr14	51096712	51096727	5.97E-10	1.08E-06	0.371	No	No
ENSG00000125375.14_4	*ATP5S*	chr14	50790662	50790878	1.54E-04	0.032180049	0.238	No	No
ENSG00000125375.14_4	*ATP5S*	chr14	50790662	50790834	1.60E-04	0.033219987	0.145	No	No
ENSG00000182979.17_5	*MTA1*	chr14	105934674	105934686	4.49E-08	3.77E-05	0.152	Yes	Yes
ENSG00000182718.16_5	*ANXA2*	chr15	60686772	60686894	1.00E-04	0.022942745	0.379	No	No
ENSG00000103888.16_2	*CEMIP*	chr15	81165878	81166037	3.10E-06	0.001342627	0.071	No	No
ENSG00000068305.17_4	*MEF2A*	chr15	100243566	100243590	2.28E-06	0.001082255	0.072	Yes	Yes
ENSG00000067141.16_2	*NEO1*	chr15	73567032	73567065	1.83E-07	1.32E-04	0.175	Yes	Yes
ENSG00000137817.16_3	*PARP6*	chr15	72541585	72541655	7.77E-16	5.22E-12	0.363	Yes	Yes
ENSG00000168096.14_3	*ANKS3*	chr16	4776659	4776781	1.55E-04	0.032356377	0.175	No	No
ENSG00000168096.14_3	*ANKS3*	chr16	4776979	4777178	1.18E-05	0.003957579	0.334	No	No
ENSG00000124074.11_2	*ENKD1*	chr16	67698898	67699071	2.05E-04	0.039612673	0.2	No	No
ENSG00000135709.12_3	*KIAA0513*	chr16	85116169	85116199	2.09E-04	0.040065546	0.13	Yes	Yes
ENSG00000008710.19_3	*PKD1*	chr16	2163041	2163066	1.46E-05	0.004687936	0.112	Yes	Partial
ENSG00000008710.19_3	*PKD1*	chr16	2163041	2163060	2.31E-04	0.042638163	0.093	Yes	Yes
ENSG00000140682.18_4	*TGFB1I1*	chr16	31485002	31485055	2.10E-04	0.040178146	0.074	No	No
ENSG00000140682.18_4	*TGFB1I1*	chr16	31485155	31485298	1.59E-04	0.032934606	0.055	No	No
ENSG00000033627.16_4	*ATP6V0A1*	chr17	40660589	40660607	0	0	0.308	Yes	Yes
ENSG00000108509.20_4	*CAMTA2*	chr17	4872794	4872815	2.52E-13	1.25E-09	0.209	Yes	Yes
ENSG00000108262.15_4	*GIT1*	chr17	27905979	27906006	2.43E-04	0.044080821	0.112	Yes	Yes

ENSG00000161647.18_3	*MPP3*	chr17	41894044	41894065	4.90E-12	1.59E-08	0.599	Yes	Yes
ENSG00000108523.15_3	*RNF167*	chr17	4843781	4844021	2.31E-04	0.042638163	0.345	No	No
ENSG00000008294.20_3	*SPAG9*	chr17	49085197	49085212	5.81E-07	3.44E-04	0.07	No	No
ENSG00000187824.8_5	*TMEM220*	chr17	10632347	10632377	2.11E-04	0.040232531	0.276	No	No
ENSG00000101639.18_4	*CEP192*	chr18	13038368	13038578	6.32E-05	0.015974124	0.397	No	No
ENSG00000168502.17_3	*MTCL1*	chr18	8779030	8779108	2.30E-04	0.042620191	0.058	No	No
ENSG00000089351.14_4	*GRAMD1A*	chr19	35513803	35513815	4.24E-13	1.99E-09	0.142	No	No
ENSG00000008441.16_5	*NFIX*	chr19	13189426	13189549	2.97E-08	2.58E-05	0.076	No	No
ENSG00000125991.19_4	*ERGIC3*	chr20	34142142	34142157	0	0	0.184	Yes	Yes
ENSG00000125772.12_3	*GPCPD1*	chr20	5566839	5566915	1.81E-05	0.005618177	0.223	No	No
ENSG00000132793.11_2	*LPIN3*	chr20	39986239	39986674	1.01E-04	0.022977137	0.239	No	No
ENSG00000132793.11_2	*LPIN3*	chr20	39986276	39986674	1.11E-04	0.024579465	0.24	No	No
ENSG00000124104.18_2	*SNX21*	chr20	44469086	44469097	2.00E-10	4.17E-07	0.401	Yes	Yes
ENSG00000101150.17_4	*TPD52L2*	chr20	62518916	62518958	0	0	0.064	Yes	Yes
ENSG00000100099.20_4	*HPS4*	chr22	26867178	26867256	1.52E-04	0.031990412	0.095	No	No
ENSG00000182287.14_5	*AP1S2*	chrX	15846314	15846323	0	0	0.151	Yes	Yes
ENSG00000147251.15_4	*DOCK11*	chrX	117819482	117819500	3.54E-07	2.27E-04	0.163	Yes	Yes
ENSG00000147121.15_3	*KRBOX4*	chrX	46331459	46331578	6.34E-05	0.015974124	0.17	No	No

Suppl. Table 2 List of 19 significantly altered exon-inclusion events (FDR<0.05 and ΔPSI >0.05) in 13 of the 111 pre-defined genes in *SRRM4*-activated patient fibroblasts compared to *SRRM4*-activated control fibroblasts (rMATS-turbo analysis[1])

Gene ID	Gene symbol	Chromosome	Exon start	Exon end	P value	FDR	Inclusion level difference	Category	Raj et al.[8] N2A [gene]	Raj et al.[8] N2A [event]	OMIM [MIM number]
ENSG00000097033.14_3	***SH3GLB1***	**chr1**	**87195770**	**87195794**	**0.001059475**	**0.032718017**	**0.092**	**High in case**	**Yes**	**Yes**	
ENSG00000189223.14_3	*PAX8-AS1*	chr2	114016840	114017161	2.80E-05	0.002038917	0.084	High in case	No	No	
ENSG00000055917.15_4	*PUM2*	chr2	20478343	20478580	0.001159442	0.034631215	0.192	High in case	No	No	
ENSG00000163697.16_4	*APBB2*	chr4	40937093	40937156	9.55E-05	0.004843029	0.081	High in case	No	No	
ENSG00000049540.16_4	*ELN*	chr7	73457447	73457498	2.80E-08	1.70E-05	0.125	High in case	No	No	123700; 185500
ENSG00000049540.16_4	*ELN*	chr7	73480023	73480062	1.24E-04	0.005941804	0.101	High in case	No	No	123700; 185500
ENSG00000198363.17_3	***ASPH***	**chr8**	**62594997**	**62595042**	**3.33E-05**	**0.002334018**	**0.1**	**High in case**	**Yes**	**Yes**	**601552**
ENSG00000165802.22_5	*NSMF*	chr9	140349690	140349759	6.78E-05	0.003743249	0.16	High in case	No	No	614838
ENSG00000110514.19_3	*MADD*	chr11	47295377	47295527	0.001631624	0.046450284	0.082	High in case	Yes	No	619004; 619005
ENSG00000182796.14_4	*TMEM198B*	chr12	56228634	56229399	0.001679533	0.047078596	0.077	High in case	No	No	
ENSG00000182796.14_4	*TMEM198B*	chr12	56228634	56228823	4.58E-04	0.016701949	0.092	High in case	No	No	
ENSG00000182718.16_5	*ANXA2*	chr15	60656627	60656722	1.41E-06	2.34E-04	0.132	High in case	No	No	
ENSG00000182718.16_5	*ANXA2*	chr15	60678226	60678285	1.86E-04	0.007889587	0.081	High in case	No	No	
ENSG00000125991.19_4	***ERGIC3***	**chr20**	**34142142**	**34142157**	**7.75E-06**	**9.20E-04**	**0.098**	**High in case**	**Yes**	**Yes**	
ENSG00000182287.14_5	***AP1S2***	**chrX**	**15846314**	**15846323**	**2.18E-07**	**7.94E-05**	**0.16**	**High in case**	**Yes**	**Yes**	**304340**
ENSG00000115414.18_4	*FN1*	chr2	216236631	216237047	9.57E-05	0.004843029	-0.059	High in CTRL	No	No	601894; 184255
ENSG00000115414.18_4	*FN1*	chr2	216257653	216257926	9.63E-08	4.39E-05	-0.142	High in CTRL	No	No	601894; 184255
ENSG00000182718.16_5	*ANXA2*	chr15	60678226	60678285	2.04E-04	0.008265056	-0.075	High in CTRL	No	No	
ENSG00000182718.16_5	*ANXA2*	chr15	60678231	60678285	2.03E-04	0.008265056	-0.081	High in CTRL	No	No	

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