Table SI. NF1 genetic variants and other cutaneous manifestations

Patient	*Gender*	*Age (years)*	*NF1 variant (Transcript: NM_001042492.3)*	*Variant /ACMG Classification**	*CALMs number*	*Mean melanin CALMs*	*Other cutaneous manifestations***
1	*F*	*11*	*Exon30 | c.4084C>T | p.Arg1362**	*P/ PVS1, PM2, PP5/ protein truncating*	*25*	*31,40*	*FR, CSN*
2	*M*	*4*	*Exon21 | c.2729del | p.Gly910fs*	*P/ PVS1, PM2, PP5/ protein truncating*	*7*	*55,67*	*FR*
3	*M*	*8,5*	*Exon29 | c.3916C>T | p.Arg1306**	*P/ PVS1, PM2, PP5/ protein truncating*	*18*	*34,00*	*FR*
4	*F*	*16*	*Exon47 | c.6943del | p.Leu2315fs*	*LP/ PVS1, PM2/ protein truncating*	*15*	*32,00*	*FR, PN, CSN*
5	*M*	*5*	*Exon32 | c.4183C>T | p.Gln1395**	*P/ PVS1, PM2, PP5/ protein truncating*	*12*	*56,27*	*FR*
6	*F*	*7,5*	*Exon31 | c.4172G>C | p.Arg1391Thr*	*LP/PM1, PP2/ non protein truncating*	*22*	*24,59*	*FR*
7	*M*	*2,5*	*Exon45 | c.6792C>A | p.Tyr2264**	*P/ PVS1, PM2, PP5/ protein truncating*	*13*	*54,67*	*PN*
8	*M*	*8*	*Exon6 | c.653dup | p.Ala219fs*	*P/ PVS1, PM2, PP5/ protein truncating*	*27*	*24,59*	*FR*
9	*F*	*5*	*Exon38 | c.5390del | p.Asn1797fs*	*P/ PVS1, PM2, PP5/ protein truncating*	*11*	*66,67*	*FR, PN*
10	*M*	*13*	*Exon19 | c.2326-3T>G*	*LP/ PM2, PP3, PP5/Splice-Altering (0.78)** / Protein truncating*	*8*	*79,67*	*FR*
11	*M*	*4*	*Exon24 | c.3142T>C | p.Trp1048Arg*	*LP/ PM2, PP3, PP2/ non protein truncating*	*20*	*80,24*	*CSN*
12	*F*	*15*	*Exon22 | c.2858T>A | p.Leu953**	*P/ PVS1, PM2, PP5/ protein truncating*	*13*	*63,10*	*FR, PN*
13	*F*	*14*	*Exon10 | c.1063-2A>G*	*P/ PVS1, PM2, PP5/ Splice-Altering (0.99)**/ Protein truncating*	*25*	*66,83*	*FR*
14	*F*	*12,5*	*Exon40 | c.5902C>T | p.Arg1968**	*P/ PVS1, PM2, PP5/ protein truncating*	*6*	*82,97*	*FR*
15	*M*	*10*	*Exon5 | c.579_581del | p.Leu194del*	*P/ PM4/ protein truncating*	*9*	*80,50*	
16	*F*	*4*	*Exon4 | c.350dup |*	*P/ PVS1, PM2, PP5/*	*28*	*74,00*	

			p.Cys118fs	*protein truncating*			
17	*M*	*3*	*Exon45 | c.6720_6721dup | p.Asn2241fs*	*P/ PVS1, PM2, PP5/ protein truncating*	*10*	*86,43*	*FR*
18	*M*	*10*	*Exon38 | c.5609G>A | p.Arg1870Gln*	*P/ PS1, PM2, PM5, PM1, PP3, PP2, PP5/ non protein truncating*	*35*	*69,40*	*FR*
19	*M*	*9*	*Exon47 | c.7000G>C | p.Gly2334Arg*	*P/ PM2, PP5/ non protein truncating*	*25*	*65,38*	*FR, CSN*
20	*F*	*6*	*Exon50 | c.7348C>T | p.Arg2450**	*P/ PVS1, PM2, PP5/ protein truncating*	*22*	*55,21*	*FR*
21	*M*	*1,5*	*Exon50 | c.7348C>T | p.Arg2450**	*P/ PVS1, PM2, PP5/ protein truncating*	*15*	*26,14*	*FR, PN*
22	*F*	*12*	*Exon38 | c.5489G>T | p.Arg1830Leu*	*P/ PM1, PP2, PP5, PM2/ non protein truncating*	*20*	*84,37*	*FR*
23	*M*	*14*	*Exon38 | c.5489G>T | p.Arg1830Leu*	*P/ PM1, PP2, PP5, PM2/ non protein truncating*	*11*	*78,81*	
24	*F*	*13*	*Exon38 | c.5609+1G>A*	*P/ PVS1, PM2, PP5/Splice-Altering (0.99)/ Protein truncating*	*14*	*76,67*	*FR*
25	*M*	*3*	*Exon40 | c.5858T>C | p.Leu1953Pro*	*P/ PP3, PM2/ non protein truncating*	*6*	*62,95*	
26	*M*	*8*	*Exon50 | c.7348C>T | p.Arg2450**	*P/ PVS1, PM2/ protein truncating*	*15*	*54,00*	*FR,PN*
27	*F*	*16*	*Exon5 | c.501T>A | p.Cys167**	*P/ PVS1, PM2, PP5/ protein truncating*	*13*	*29,03*	*FR*
28	*F*	*2*	*Exon13 | c.1466A>G | p.Tyr489Cys*	*P/ PM2, PM1, PP2/ non protein truncating*	*24*	*61,07*	*FR*
29	*F*	*13*	*Exon21 | c.2693T>C | p.Leu898Pro*	*P/ PP3, PM2/ non protein truncating*	*36*	*94,27*	*FR, JXG*
30	*M*	*2,5*	*Exon46 | c.6855C>A | p.Tyr2285**	*P/ PVS1, PM2, PP5/ protein truncating*	*9*	*50,82*	
31	*M*	*11*	*Exon15 | c.1691_1692insC | p.Ala565fs*	*P/ PVS1, PM2, PP5/ protein truncating*	*18*	*98,00*	*FR, PN, NA*
32	*M*	*10*	*Exon18 | c.2248dup | p.Thr750fs*	*P/ PVS1, PM2, PP5/ protein truncating*	*11*	*96,52*	*FR,PN*
33	*M*	*4*	*Exon38 | c.5352T>G | p.Tyr1784**	*LP/ PVS1, PM2/ protein truncating*	*18*	*98,07*	*FR,PN*
34	*M*	*6*	*Exon32 | c.4331A>G | p.Lys1444Arg*	*P/ PM1, PP2, PM2/ non protein truncating*	*13*	*112,09*	*FR*

35	*F*	*9*	*Exon9 | c.1021_1022del | p.Val341fs*	*P/ PVS1, PM2, PP5/ protein truncating*	*17*	*87,26*	*FR,PN*
36	*F*	*1,75*	*Exon32 | c.4331A>G | p.Lys1444Arg*	*P/ PM1, PP2, PM2/ non protein truncating*	*21*	*39,16*	*FR*
37	*M*	*4*	*Exon52 | c.7739-3C>A*	*VUS/ PM2, PP3/ Splice-Altering (low) (0.44)/ non protein truncating*	*22*	*88,22*	*FR*
38	*F*	*11*	*Exon22 | c.2970_2972del | p.Met991del*	*P/ PM4, PM5/ protein truncating*	*8*	*133,00*	
39	*F*	*10*	*Exon22 | c.2970_2972del | p.Met991del*	*P/ PM4, PM5/ protein truncating*	*16*	*98,74*	*PN*
40	*F*	*8*	*Exon22 | c.2970_2972del | p.Met991del*	*P/ PM4, PM5/ protein truncating*	*13*	*106,71*	*JXG*
41	*F*	*16*	*Exon34 | c.4578-9T>A*	*VUS/ PM2, PP3/ Splice-Altering (0.64)/ non protein truncating*	*13*	*88,00*	*CSN*
42	*M*	*4*	*Exon18 | c.2033dup | p.Ile679fs*	*P/ PVS1, PM2, PP5/ protein truncating*	*10*	*42,67*	*PN*
43	*M*	*16*	*Exon9 | c.1022_1023insGA | p.Ile342fs*	*P/ PVS1, PM2, PP5/ protein truncating*	*37*	*74,91*	*FR,PN,CSN*
44	*M*	*9*	*Exon7 | c.693del | p.Phe231fs*	*P/ PVS1, PM2, PP5/ protein truncating*	*16*	*75,23*	*FR*
45	*M*	*9*	*Exon38 | c.5488C>T | p.Arg1830Cys*	*P/ PM1, PP2, PM2/ non protein truncating*	*13*	*78,83*	*FR*
46	*F*	*2*	*Exon25 | c.3198-2A>G*	*P/ PVS1, PM2, PP5/ Splice-Altering (0.97)/ protein truncating*	*28*	*55,43*	*FR*
47	*F*	*0,58*	*Exon17 | c.1920del | p.Ser641fs*	*P/ PVS1, PM2, PP5/ protein truncating*	*25*	*35,54*	*PN*
48	*M*	*11*	*Exon35 | c.4613del | p.Lys1538fs*	*LP/ PVS1, PM2/ protein truncating*	*23*	*43,97*	*FR, PN*
49	*F*	*5*	*Exon34 | c.4432_4433insAA | p.Phe1478fs*	*P/ PVS1, PM2, PP5/ protein truncating*	*35*	*75,76*	*FR*
50	*F*	*3*	*Exon21 | c.2540T>C | p.Leu847Pro*	*P/ PM1, PP2/ non protein truncating*	*23*	*79,20*	*JXG*
51	*M*	*1*	*Exon30 | c.4084C>T | p.Arg1362**	*P/ PVS1, PM2, PP5/ protein truncating*	*25*	*42,76*	
52	*F*	*2,5*	*Exon21 | c.2729del |*	*P/ PVS1, PM2, PP5/ protein*	*7*	*35,58*	

			p.Gly910fs	*truncating*			
53	*M*	*9*	*Exon29 | c.3916C>T | p.Arg1306**	*P/ PVS1, PM2, PP5/ protein truncating*	*18*	*93,63*	*PN, CSN*
54	*M*	*13*	*Exon47 | c.6943del | p.Leu2315fs*	*LP/PVS1, PM2/ protein truncating*	*15*	*52,93*	
55	*F*	*0,5*	*Exon32 | c.4183C>T | p.Gln1395**	*P/ PVS1, PM2, PP5/ protein truncating*	*12*	*57,10*	
56	*F*	*11*	*Exon32 | c.4235G>C | p.Arg1412Thr*	*LP/PM1, PP2/ non protein truncating*	*22*	*70,71*	*FR, PN, NA, CSN*
57	*F*	*1,25*	*Exon46 | c.6855C>A | p.Tyr2285**	*P/ PVS1, PM2, PP5/ protein truncating*	*13*	*57,28*	
58	*M*	*3*	*Exon6 | c.653dup | p.Ala219fs*	*P/ PVS1, PM2, PP5/ protein truncating*	*27*	*31,40*	*FR*
59	*M*	*3*	*Exon26 | c.3347_3350del | p.Asp1116fs*	*P/PVS1, PM2, PP5/ protein truncating*			*FR*
60	*F*	*5*	*Exon 29 | c.3942G>A, p.Trp1314**	*P/PVS1, PM2, PP5/ protein truncating*			*FR*
61	*M*	*4,5*	*Exon3| c.288_288+2del | p.Gln97fs*	*P/ PVS1, PM2, PP5/ Splice-Altering (0.98)/ protein truncating*			*JXG*
62	*M*	*7*	*Exon28 |c.3789del | p.Glu1264fs*	*P/ PVS1, PM2, PP5/ protein truncating*			*FR,CSN*
63	*F*	*13*	*Exon19 | c.2326-3T>G |*	*LP (Likely pathogenic/ PM2, PP3, PP5/ Splice-Altering (0.78)**/ Truncating mutation*			*FR*

Abbreviations: *F Female, M Male, CALMs Cafe-au-lait macules, FR freckling, CSN cutaneous/subcutaneous neurofibromas, PN plexiform neurofibromas, JXG juvenile xanthogranulomas, NA nevus anemicus

***** SpliceAI uses deep neural networks to predict whether splicing events occur. The score can range from 0 to 1, when scores can be interpreted as the probability of the variant being splice-altering.***