

Supplementary Table 3 NF1 germline and somatic mutations.
 Supplementary Table 4 NF1 germline and somatic mutations identified in patients with matched blood DNA available.

NF1 GERMLINE MUTATIONS																													
Case ID	Type	Chr	Start	End	Ref	Alt	Function	Effect	Normal VAF	Tumor VAF	Tumor VAF (normalized by purity)	Tumor Genotype	Sanger Validation	ClinVar ID	HGMD ID	Pathogenicity	Missense Pathogenic Predictions	MutFaster missense	PolyPhen2 missense	Provar missense	SIFT missense	In-frame indel Pathogenic Predictions	FATHMM in-frame indel	Provar in-frame indel	SIFT in-frame indel	VEST in-frame indel	Log odds ratio 1		
1	DEL	17	29527228	29527228	C	T	splicing	p.29527228C>T	0.0	0.0	0.0	DN	verified																
4	DEL	17	29546266	29546267	AG	T	frameshift	p.29546266G>T	0.43	0.43	1.00	DN	verified	14817026500	CS662397	frameshift												58.64	
5	DEL	17	29545141	29545146	AGAC	T	frameshift	p.29545141G>T	0.0	0.0	0.0	DN	verified			frameshift												58.64	
6	DEL	17	29564869	29564869	C	T	frameshift	p.29564869C>T	0.0	0.0	0.0	DN	verified			frameshift												58.64	
7	SNV	17	29552277	29552277	G	C	splicing	p.29552277G>C	0.46	0.4	1.00	DN	verified		CS600875	splicing												92.67	
9	SNV	17	29616116	29616116	A	G	splicing	p.29616116A>G	0.46	0.38	1.00	DN	verified		CS600874	splicing												92.76	
10	SNV	17	29678364	29678364	C	T	frameshift	p.29678364C>T	0.75	1.00	1.00	DN	verified	14865446137	CS641376	frameshift												45.82	
11	DEL	17	29494344	29494347	TCTT	T	frameshift	p.29494344T>C	0.0	0.0	0.0	DN	verified	13786258474	CS600348	frameshift												3.79	
12	DEL	17	29546048	29546048	C	T	frameshift	p.29546048C>T	0.0	0.0	0.0	DN	verified			frameshift												not available	
13	DEL	17	29551544	29551544	T	T	frameshift	p.29551544T>T	0.0	0.0	0.0	DN	verified			frameshift												3.4	
15	DEL	17	29543332	29543332	AA	T	frameshift	p.29543332A>T	0.23	0.23	1.00	DN	verified			frameshift												11.63	
18	DEL	17	29544844	29544844	TAC	T	frameshift	p.29544844T>C	0.23	0.23	1.00	DN	verified	13786258781	CS621129	frameshift												19.81	
19	SNV	17	29509171	29509171	T	T	frameshift	p.29509171T>T	0.13	0.13	1.00	DN	verified	13786259305	CS641379	frameshift												49.21	
20	SNV	17	2954447	2954447	C	T	frameshift	p.2954447C>T	0.0	0.0	1.00	DN	verified	1481738407		frameshift												87.21	
21	SNV	17	2954118	2954118	A	G	splicing	p.2954118A>G	0.43	0.24	1.00	DN	verified			frameshift												49.21	
21	SNV	17	29588761	29588761	C	T	frameshift	p.29588761C>T	0.43	0.0	1.00	DN	verified	13786259305	CS641379	frameshift												6.83	
22	SNV	17	29494028	29494028	T	T	splicing	p.29494028T>T	0.0	0.0	1.00	DN	verified			splicing												28.52	
23	SNV	17	29505444	29505444	G	T	frameshift	p.29505444G>T	0.45	0.45	1.00	DN	verified			frameshift												35.79	
27	DEL	17	29543831	29543831	T	T	frameshift	p.29543831T>T	0.43	0.43	1.00	DN	verified			frameshift												23.29	
28	SNV	17	29528178	29528178	G	A	splicing	p.29528178G>A	0.39	0.38	1.00	DN	verified	14864127261	CS600650	splicing												28.23	
29	DEL	17	2954448	2954448	T	T	frameshift	p.2954448T>T	0.43	0.43	1.00	DN	verified			frameshift												18.0	
30	INS	17	29583369	29583369	AG	T	splicing	p.29583369AG>T	0.43	0.43	1.00	DN	verified	13786259305	CS600651	splicing												51.76	
31	SNV	17	29585011	29585011	C	T	frameshift	p.29585011C>T	0.43	0.43	1.00	DN	verified		CS641385	frameshift												61.09	
39	DEL	17	2959368	2959368	T	T	frameshift	p.2959368T>T	0.13	0.13	1.00	DN	verified			frameshift												18.5	
42	SNV	17	29598778	29598778	C	A	frameshift	p.29598778C>A	0.47	0.47	1.00	DN	verified		CS686301	frameshift												37.20	
43	SNV	17	2959524	2959524	T	A	frameshift	p.2959524T>A	0.29	0.29	1.00	DN	verified			frameshift												49.01	
44	DEL	17	2966763	2966763	ACTT	T	frameshift	p.2966763ACTT>T	0.43	0.43	1.00	DN	verified	1486324848	CS611709	frameshift												57.60	
46	SNV	17	2959596	2959596	A	A	frameshift	p.2959596A>A	0.2	0.2	1.00	DN	verified	14864747174	CS686304	frameshift												193.66	
47	SNV	17	2954545	2954545	A	A	frameshift	p.2954545A>A	0.43	0.43	1.00	DN	verified		CS600789	frameshift												17.66	
47*	SNV	17	2954683	2954683	TCT	T	in-frame	p.2954683TCT>TCT	0.4	0.4	1.00	DN	verified			pathogenic in-frame indel							3/4					17.66	
48	SNV	17	2987502	2987502	T	C	missense	p.2987502T>C	0.54	0.54	1.00	DN	verified	CS654014		pathogenic missense												67.17	
49	SNV	17	2954070	2954070	T	T	frameshift	p.2954070T>T	0.38	0.38	1.00	DN	verified		CS611468	frameshift												11.45	
51	DEL	17	2956029	2956029	AA	T	frameshift	p.2956029AA>T	0.29	0.29	1.00	DN	verified			frameshift												18.0	
52*	SNV	17	29527438	29527438	A	G	splicing	p.29527438A>G	0.48	0.31	0.61	NS	verified	1487883922	CS691467	splicing												0.00	
52*	SNV	17	2969279	2969279	C	T	missense	p.2969279C>T	0.13	0.13	0.61	NS	verified		CS64442	splicing													72.81
53	SNV	17	29528204	29528204	G	T	splicing	p.29528204G>T	0.13	0.13	1.00	DN	verified		CS672951	frameshift												160.68	
56	DEL	17	2951818	2951818	TCTC	T	frameshift	p.2951818TCTC>T	0.1	0.1	0.61	NS	verified			frameshift												not available	
57	SNV	17	2931378	2931378	C	T	frameshift	p.2931378C>T	0.47	0.51	0.91	DN	verified	1487883985	CS600780	frameshift												17.66	
66	SNV	17	2956545	2956545	A	G	splicing	p.2956545A>G	0.13	0.0	0.61	NS	verified			splicing												17.66	
67	DEL	17	2950593	2950593	GG	T	frameshift	p.2950593GG>T	0.43	0.43	1.00	DN	verified			frameshift												0.00	

NF1 SOMATIC MUTATIONS																											
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6	DEL	17	29552277	29552277	C	T	frameshift	p.29552277C>T	0.0	verified			frameshift														1.00
14	SNV	17	2955482	2955482	C	A	missense	p.2955482C>A	0.18	verified			pathogenic missense	4/4													3.77
17	INS	17	2956207	2956207	T	A	frameshift	p.2956207T>A	0.29	verified	1486041348		frameshift														12.4
20	INS	17	2956563	2956563	TCTGT	T	in-frame	p.2956563TCTGT>T	0.29	verified			pathogenic in-frame indel														8.00
52	SNV	17	2951480	2951480	T	T	frameshift	p.2951480T>T	0.46	verified	1374678261		frameshift														0.00
55	DEL	17	2956480	2956480	T	T	frameshift	p.2956480T>T	0.1	verified			frameshift														not available
66	SNV	17	2956469	2956469	C	T	frameshift	p.2956469C>T	0.28	verified			frameshift														17.66
67	SNV	17	2952744	2952744	T	C	splicing	p.2952744T>C	0.34	verified		1374678261	CS601246	frameshift													not available

*Samples with two independent NF1 germline mutations.