

Description	Ref	Exp	cell type	Family#	Member#	Relationship	Age	Unit	Gender	Race	Ethnicity	Gene	Allele#	Gene Mutation	Catalog Remark
NF1	GM01633	0	Fibroblast	113	1	proband	61	YR	M	CAUC					Atypical; see GM01634 Lymphoid
NF1	GM01634	0	B-Lymphocyte	113	1	proband	61	YR	M	CAUC					Atypical; see GM01633 Fibroblast
NF1	GM01639	0	Fibroblast	114	1	proband	19	YR	F	BL					Normal skin; other fibroblasts also available; see GM01641 Lymphoid
NF1	GM01641	0	B-Lymphocyte	114	1	proband	19	YR	F	BL					Clinically affected; see GM01639 Fibro
NF1	GM01858	B	Fibroblast	115	1	proband	41	YR	M	CAUC					Neurofibroma biopsy-fibroblast culture; see GM01861A Lymphoid
NF1	GM01859	0	Fibroblast	115	1	proband	41	YR	M	CAUC					Cafe au lait spot biopsy-fibroblast culture; same patient as GM01858B; see GM01861A Lymphoid
NF1	GM01861	A	B-Lymphocyte	115	1	proband	41	YR	M	CAUC					HLA type A3,A2,B27,Bw22(Bw40); see also GM01858B and GM01859 Fibroblast; cafe au lait spots and neurofibromas
NF1	GM03421	0	B-Lymphocyte	212	1	affected mother	39	YR	F	CAUC					Multiple cutaneous neurofibromas; positive family history
NF1	GM03420	0	B-Lymphocyte	212	2	proband	17	YR	F	CAUC					Cafe au lait spots; neurofibromas; mental retardation; positive family history
NF2	GM09305	0	B-Lymphocyte	2148	1	proband			M						A son, 2 brothers, 1 sister, & father are also affected
NF2	GM09306	A	B-Lymphocyte	2148	3	son	27	YR	M	CAUC					Clinically unaffected; father & sister are affected
NF2	GM09307	A	B-Lymphocyte	2148	4	affected son			M						Clinically affected; father is also affected
NF1	GM09534	0	B-Lymphocyte	2176	1	proband	47	YR	F	CAUC	IRISH/ SCOTTISH/ ENGLISH				Neurofibromas; café au lait spots; mother, 4 sibs, a daughter, and a granddaughter are also affected; 46,XX
NF1	GM09535	A	B-Lymphocyte	2176	3	affected granddaughter	6	YR	F	CAUC	IRISH/ SCOTTISH/ ENGLISH				Multiple cafe au lait spots; slightly slower development than sibs; mother and grandmother are affected; sister of GM09539

Description	Ref	Exp	cell type	Family#	Member#	Relationship	Age	Unit	Gender	Race	Ethnicity	Gene	Allele#	Gene Mutation	Catalog Remark
NF1	GM09539	0	B-Lymphocyte	2176	4	granddaughter	5	YR	F	CAUC	IRISH/ SCOTTISH/ ENGLISH ITALIAN/ ENGLISH			At risk; clinically asymptomatic; mother, sister, and grandmother are affected	
NF1	GM09536	0	B-Lymphocyte	2176	6	in-law	54	YR	F	CAUC				Unaffected spouse of GM09537	
NF1	GM09616	0	B-Lymphocyte	2176	7	affected sister	55	YR	F	CAUC				Multiple neurofibromas; multiple café au lait spots; 1 affected son	
NF1	GM09619	0	B-Lymphocyte	2176	8	affected nephew	26	YR	M	CAUC				Multiple neurofibromas; multiple café au lait spots; pseudoarthrosis; large spinal tumor; pathologically verified; son of GM09616; mother is also affected	
NF1	GM09617	0	B-Lymphocyte	2176	9	niece	28	YR	F	CAUC				Clinically unaffected; mother and 4 sibs are affected; daughter of GM09627	
NF1	GM09618	A	B-Lymphocyte	2176	10	in-law	31	YR	M	CAUC				Clinically unaffected; spouse of GM09617	
NF1	GM09620	0	B-Lymphocyte	2176	11	niece	39	YR	F	CAUC				Clinically unaffected; grandmother, 3 maternal aunts & 2 maternal uncles are affected	
NF1	GM09621	0	B-Lymphocyte	2176	12	great nephew	8	YR	M	CAUC				Clinically unaffected; son of GM09617	
NF1	GM09622	0	B-Lymphocyte	2176	13	great nephew	7	YR	M	CAUC				Clinically unaffected; son of GM09617	
NF1	GM09625	0	B-Lymphocyte	2176	14	maternal uncle	67	YR	M	CAUC				Clinically unaffected; 2 affected sibs; father was affected	
NF1	GM09626	0	B-Lymphocyte	2176	15	in-law	62	YR	F	CAUC				Clinically unaffected; spouse of GM09625	
NF1	GM09627	0	B-Lymphocyte	2176	16	affected sister	52	YR	F	CAUC				Multiple neurofibromas; optic glioma; mother, 4 sibs, and 4 children are affected	
NF1	GM09628	0	B-Lymphocyte	2176	17	affected brother	41	YR	M	CAUC				Multiple neurofibromas; multiple café au lait spots; 3 affected children	
NF1	GM09629	0	B-Lymphocyte	2176	18	in-law	32	YR	F	CAUC				Clinically unaffected; spouse of GM09628; 1 affected child	
NF1	GM09630	0	B-Lymphocyte	2176	19	maternal cousin	32	YR	M	CAUC				Clinically unaffected; son of GM09625	
NF1	GM09631	0	B-Lymphocyte	2176	20	maternal cousin	29	YR	F	CAUC				Clinically unaffected; daughter of GM09625	

Description	Ref	Exp	cell type	Family#	Member#	Relationship	Age	Unit	Gender	Race	Ethnicity	Gene	Allele#	Gene Mutation	Catalog Remark
NF1	GM09632	0	B-Lymphocyte	2176	21	maternal cousin	33	YR	F	CAUC					Clinically unaffected; daughter of GM09625
NF1	GM09633	0	B-Lymphocyte	2176	22	affected niece	16	YR	F	CAUC					Multiple cafe au lait spots; daughter of GM09627
NF1	GM09634	0	B-Lymphocyte	2176	23	affected nephew	15	YR	M	CAUC					Multiple cafe au lait spots; son of GM09627
NF1	GM09635	0	B-Lymphocyte	2176	24	nephew	35	YR	M	CAUC					5 fatty nodules reportedly lipomas; son of GM09616; mother and brother are affected
NF1	GM09636	0	B-Lymphocyte	2176	25	in-law	35	YR	F	CAUC					Clinically unaffected; spouse of GM09635
NF1	GM09637	A	B-Lymphocyte	2176	26	affected nephew	7	MO	M	CAUC					Multiple cafe au lait spots; son of GM09628 and GM09629; father and 2 halfbrothers are also affected
NF1	GM09638	0	B-Lymphocyte	2176	27	niece	29	YR	F	CAUC					Clinically unaffected; daughter of GM09537; paternal grandmother was affected
NF1	GM09639	0	B-Lymphocyte	2176	28	niece	25	YR	F	CAUC					Clinically unaffected; daughter of GM09537; paternal grandmother was affected
NF1	GM09640	0	B-Lymphocyte	2176	29	grandniece	17	YR	F	CAUC					Clinically unaffected; daughter of GM09635; pat grandmother is affected
NF1	GM09641	A	B-Lymphocyte	2176	30	grandniece	10	YR	F	CAUC					Clinically unaffected; daughter of GM09635; pat grandmother is affected
NF1	GM09642	0	B-Lymphocyte	2176	31	great nephew	7	YR	M	CAUC					Clinically unaffected; son of GM09635; paternal grandmother is affected
NF1	GM09649	A	B-Lymphocyte	2176	32	in-law	67	YR	F	CAUC					Clinically unaffected; spouse was unaffected maternal uncle of proband
NF1	GM09650	0	B-Lymphocyte	2176	33	maternal cousin	49	YR	F	CAUC					Clinically unaffected; daughter of GM09649; pat grandfather was affected
NF1	GM09651	0	B-Lymphocyte	2176	34	maternal cousin	38	YR	M	CAUC					Clinically unaffected; son of GM09649; paternal grandfather was affected
NF1	GM09652	0	B-Lymphocyte	2176	35	maternal cousin	39	YR	F	CAUC					Clinically unaffected; daughter of GM09649; paternal grandfather was affected
NF1	GM09688	A	B-Lymphocyte	2176	36	maternal cousin			M	CAUC					Clinically unaffected; father, brother (GM09692A), and niece (GM09690A) are affected; son of GM09691
NF1	GM09692	A	B-Lymphocyte	2176	37	affected maternal cousin			M	CAUC					Multiple neurofibromas; facial paralysis due to neurofibromas and surgery; father and daughter (GM09690A) are also affected; spouse of GM09689; son of GM09691

Description	Ref	Exp	cell type	Family#	Member#	Relationship	Age	Unit	Gender	Race	Ethnicity	Gene	Allele#	Gene Mutation	Catalog Remark
NF1	GM09689	B	B-Lymphocyte	2176	38	in-law			F	CAUC					Spouse of GM09692A; clinically unaffected; spouse & daughter (GM09690A) are affected
NF1	GM09690	A	B-Lymphocyte	2176	39	affected maternal cousin			F	CAUC					Diagnosed at age 17 months; neurofibroma in throat at age 3; multiple neurofibromas; scoliosis surgery; malignancy in larynx; daughter of GM09689 and GM09692A
NF1	GM09691	0	B-Lymphocyte	2176	40	in-law			F	CAUC					Clinically unaffected mother of GM09692A and GM09688A
NF1	GM09693	0	B-Lymphocyte	2176	41	affected brother	51	YR	M	CAUC					Multiple neurofibromas; cafe au lait spots; grandfather, mother, and 4 sibs are also affected
NF1	GM09694	0	B-Lymphocyte	2176	42	grandniece	8	YR	F	CAUC					Clinically unaffected; father is affected; affected paternal grandmother is GM09616
NF1	GM09886	A	B-Lymphocyte	2176	43	maternal aunt	74	YR	F	CAUC					Clinically unaffected; father & 2 sibs are affected
NF1	GM09887	A	B-Lymphocyte	2176	44	sister			F	CAUC					Clinically unaffected; mother and 5 sibs are affected; mother of GM09620
NF1	GM09944	0	B-Lymphocyte	2176	45	nephew	33	YR	M	CAUC					Clinically unaffected with a few cafe au lait spots and lipomas; father of GM09694; son of GM09616; mother and brother are affected
NF1	GM00622	A	Fibroblast			proband	8	YR	M						Clinically affected
NF1	GM11601	0	B-Lymphocyte			proband	31	YR	M	CAUC		NF1	1	ALU INS	Multiple cervical nerve root neurofibromas; 1 peripheral neurofibroma; macrocephaly; Lisch nodules; no cafe au lait spots or freckling; de novo insertion of an Alu repetitive element close to exon 6 of NF1 gene leading to abnormal gene splicing
NF1	GM11602	0	B-Lymphocyte			proband	11	YR	F	CAUC		NF1	1	1-BP DEL, EX 1	Multiple cafe au lait spots; axillary freckling; acute lymphocytic leukemia in remission; deletion of a single nucleotide in 'exon 1' of the NF1 gene leading to a frameshift mutation