

SUPPLEMENTARY TABLES

ID	Sex	Age	Clinical Features	Clinical Group
NF_2	F	34	<ul style="list-style-type: none"> - CALMs - Cutaneous neurofibromas - Axillary freckling - Lisch nodules 	G1
NF_54	F	47	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - UBOs 	G1
NF_68	M	35	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Spinal neurofibromas 	G1
NF_83	F	70	<ul style="list-style-type: none"> - CALMs - Cutaneous neurofibromas 	G1
NF_89	F	56	<ul style="list-style-type: none"> - CALMs - Subcutaneous fibromas 	G1
NF_1	M	51	<ul style="list-style-type: none"> - CALMs - Cutaneous neurofibromas - Axillary and inguinal freckling - Spine deformities (scoliosis) - Learning disabilities - Anxiety disorder 	G2
NF_3	M	65	<ul style="list-style-type: none"> - Cutaneous neurofibromas - Axillary and inguinal freckling - Cardiovascular disorder (hypertension) - Obesity - Hepatic steatosis - Spine (scoliosis) and bone deformities (pes cavus) 	G2

NF_4	F	35	<ul style="list-style-type: none"> - CALMs - Cutaneous neurofibromas - Axillary freckling - Spine deformities (scoliosis) - Bone deformities (bilateral pes cavus) 	G2
NF_5	F	44	<ul style="list-style-type: none"> - CALMs - Subcutaneous neurofibromas - Axillary freckling - Spine deformities (scoliosis) - Anxiety disorder - Cardiovascular disorders (hypertension) 	G2
NF_8	M	33	<ul style="list-style-type: none"> - Spinal neurofibromas - Spine deformities (scoliosis) - Learning disabilities - Obesity 	G2
NF_10	F	26	<ul style="list-style-type: none"> - CALMs - Subcutaneous neurofibromas - Spine (scoliosis) and bone (dural ectasia) deformities - Cardiovascular disorders (hypertension) - Learning disabilities 	G2
NF_11	F	59	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Spine (joint spondylosis) and bone (osteoporosis) deformities - Anxiety disorder - Sleep disorder - Cardiovascular disorders (mitral and aortic insufficiency) 	G2
NF_13	F	33	<ul style="list-style-type: none"> - CALMs - Cutaneous neurofibromas - Axillary freckling - Lisch nodules - UBOs - Spine deformities (scoliosis) - Obesity 	G2
NF_20	M	18	<ul style="list-style-type: none"> - Cutaneous neurofibromas - Inguinal freckling - Lisch nodules - Spine (kyphoscoliosis) and bone (foot deformities) deformities - Learning disabilities 	G2
NF_21	F	28	<ul style="list-style-type: none"> - CALMs - Subcutaneous neurofibromas - Spine (scoliosis, hydromyelia) and bone (short stature) deformities 	G2

			<ul style="list-style-type: none"> - Cardiovascular disorders (mitral insufficiency) - Plexiform neurofibroma in the right medial malleolus 	
NF_25	F	26	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Axillary and inguinal freckling - Bone deformities (short stature) - Hearing loss - Learning disabilities 	G2
NF_28	F	41	<ul style="list-style-type: none"> - CALMs - Cutaneous neurofibromas - Spine deformities (lost of the normal cervical lordosis) - Obesity - 25-OH Vitamin D deficiency 	G2
NF_30	F	45	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Spinal neurofibromas - Axillary and inguinal freckling - Spine and bone (short stature) deformities - Cardiovascular disorder (hypertension) - UBOs 	G2
NF_38	M	27	<ul style="list-style-type: none"> - CALMs - Behavior disorders - Spine (scoliosis) and bone (short stature) deformities 	G2
NF_40	F	41	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Lisch nodules - Vitamin D 25-OH deficiency - Microcytic anemia - Hearing loss - Spine deformities (kyphoscoliosis) - Fibrocystic mastopathy - Learning disabilities 	G2
NF_43	F	30	<ul style="list-style-type: none"> - CALMs - Subcutaneous neurofibromas - Spine deformities (scoliosis) - Cardiovascular disorder (hypertension) - Anxiety disorder - Breast lump - UBOs 	G2
NF_45	M	70	<ul style="list-style-type: none"> - CALMs - Subcutaneous neurofibromas 	G2

			<ul style="list-style-type: none"> - Epilepsy syndrome - Hypercholesterolemia - Hearing loss - Cardiovascular disorders (hypertension) 	
NF_46	M	39	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Spinal neurofibromas - Plexiform neurofibromas of the right sheath - Spine deformities (scoliosis) 	G2
NF_47	M	36	<ul style="list-style-type: none"> - CALMs - Subcutaneous neurofibromas - Inguinal freckling - Drug resistant epilepsy syndrome - Spine deformities (scoliosis) 	G2
NF_48	F	39	<ul style="list-style-type: none"> - CALMs - Subcutaneous neurofibromas - Axillary freckling - UBOs 	G2
NF_50	M	58	<ul style="list-style-type: none"> - CALMs - Cutaneous neurofibromas - Axillary and inguinal freckling - Spine deformities (scoliosis) 	G2
NF_51	F	29	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Spinal neurofibromas - Plexiform neurofibroma - UBOs - Erythrocythemia - Spine (disc protrusions) and bone (short stature) deformities - 25-OH Vitamin D deficiency - Anxiety disorder 	G2
NF_53	F	23	<ul style="list-style-type: none"> - CALMs - Spine deformities (scoliosis) - Cardiovascular disorder (coarctation of the aorta) - Anxiety disorder - Learning disabilities 	G2
NF_56	M	56	<ul style="list-style-type: none"> - CALMs - Subcutaneous neurofibromas - Spine (scoliosis) and bone deformities - Cardiovascular disorder (hypertension) - Gastric disorder 	G2

			- Multiple hepatic cysts	
NF_57	M	37	- CALMs - Cutaneous neurofibromas - Spine (kyphoscoliosis) and bone (short stature) deformities	G2
NF_59	M	52	- CALMs - Cutaneous and subcutaneous neurofibromas - Inguinal freckling - Lisch nodules - Spine (scoliosis) and bone (pectus carinatum) deformities - Cardiovascular disorders (hypertension)	G2
NF_61	F	59	- Spine (scoliosis) and bone (dural ectasia) deformities - Anxiety and depressive disorder	G2
NF_62	F	32	- CALMs - Upper limb plexiform neurofibroma - Spine deformities (scoliosis) - Anxiety disorder - D25-OH Vitamin deficiency	G2
NF_63	F	54	- CALMs - Cutaneous neurofibromas - Axillary and inguinal freckling - Lisch nodules - Spine deformities (scoliosis) - Hypercholesterolemia	G2
NF_66	M	63	- CALMs - Subcutaneous neurofibromas - Spinal neurofibromas - Spine deformities (joint spondylosis) - Cardiovascular disorders (hypertension)	G2
NF_71	F	40	- CALMs - Cutaneous and subcutaneous neurofibromas - Plexiform neurofibroma of the buttock region - Spine deformities (scoliosis) - Anxiety disorder - 25-OH Vitamin D deficiency	G2

NF_77	M	20	<ul style="list-style-type: none"> - CALMs - Cutaneous neurofibromas - Axillary and inguinal freckling - Spine (scoliosis) and bone (dural ectasia) deformities - Cardiovascular disorders (mild tricuspid valve insufficiency) 	G2
NF_80	M	32	<ul style="list-style-type: none"> - Cutaneous neurofibromas - Lisch nodules - UBOs 	G2
NF_86	F	57	<ul style="list-style-type: none"> - CALMs - Spine (scoliosis) and bone (osteoporosis) deformities 	G2
NF_90	F	26	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Spine (scoliosis) and bone deformities (foot deformities) - UBOs 	G2
NF_94	M	59	<ul style="list-style-type: none"> - CALMs - Ischemic cerebrovascular disease - Bone deformities (short stature) - Mental retardation - Glaucoma 	G2
NF_96	F	70	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Spine (scoliosis and joint spondylosis) and bone deformities (osteopenia and osteoporosis) - Cardiovascular disorder (hypertension) - 25-OH Vitamin D deficiency 	G2
NF_100	F	57	<ul style="list-style-type: none"> - CALMs - Cutaneous neurofibromas - Spine deformities (joint spondylosis) - Cardiovascular disorders (hypertension) - Anxiety and depressive disorders - Hepatic steatosis 	G2
NF_102	M	33	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Spine (kyphosis) and bone deformities - UBOs 	G2
NF_103	M	24	<ul style="list-style-type: none"> - CALMs - Spine deformities (scoliosis) - Freckling - Lisch nodules 	G2

			<ul style="list-style-type: none"> - Neurofibromas - Thorax abnormalities 	
NF_18	F	22	<ul style="list-style-type: none"> - Lisch nodules - Spine (scoliosis) and bone deformities (non-ossifying fibroma of tibia) - UBOs - Learning disabilities - Lumbar spinal MPNST 	G3
NF_52	M	46	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - MPNST of the left limb - GIST 	G3
NF_127	M	19	<ul style="list-style-type: none"> - Intracranial MPNST 	G3
NF_6	F	33	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Inguinal freckling - Spine (scoliosis) and bone (osteoporosis) deformities - Cardiovascular disorders (heart murmur) - Optic glioma 	G4
NF_17	F	26	<ul style="list-style-type: none"> - CALMs - Cutaneous neurofibromas - Axillary and inguinal freckling - Bone deformities (short stature) - Spinal optic neurofibroma - Malignant tumor at sacral region 	G4
NF_26	F	35	<ul style="list-style-type: none"> - CALMs - Subcutaneous neurofibromas - Spinal neurofibromas - Lisch nodules - Spine (scoliosis) and bone (dural ectasia) deformities - Multiple intraforaminal and paravertebral plexiform neurofibromas - UBOs - Anxiety disorder - Pilocytic astrocytoma 	G4
NF_35	M	55	<ul style="list-style-type: none"> - Cutaneous neurofibromas - Orbito-temporal plexiform neurofibroma - Chondropathy - Diverticulosis - 25-OH Vitamin D deficiency - Spinal neurinoma 	G4
NF_36	M	42	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas 	G4

			<ul style="list-style-type: none"> - Axillary and inguinal freckling - Spine deformities (scoliosis) - Spinal neuromas 	
NF_37	M	23	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Spinal neurofibromas - Spine deformities (scoliosis) - Bilateral thalamic UBOs - C2 ganglioneuroma 	G4
NF_41	F	58	<ul style="list-style-type: none"> - CALMs - Cutaneous neurofibromas - Axillary and inguinal freckling - Spine deformities (scoliosis) - Hearing loss - Cardiovascular disorders (hypertension) - Optic glioma - MGUS 	G4
NF_60	F	42	<ul style="list-style-type: none"> - CALMs - Subcutaneous neurofibromas - Spine (scoliosis) and bone (short stature) deformities - UBOs - 25-OH Vitamin D deficiency - Astrocytoma 	G4
NF_65	F	30	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Spine deformities (scoliosis) - Hearing loss - Learning disabilities - Cardiovascular disorder (hypertension) - Optic glioma 	G4
NF_67	M	52	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Spinal neurofibromas - Severe spastic tetraparesis - Spinal astrocytoma 	G4
NF_70	F	49	<ul style="list-style-type: none"> - Cutaneous and subcutaneous neurofibromas - Spine deformities (scoliosis) - Anxiety disorder - Cardiovascular disorder (heart murmur) - Microcytic anemia 	G4

			<ul style="list-style-type: none"> - Vitamin D 25-OH deficiency - Optic glioma 	
NF_72	M	38	<ul style="list-style-type: none"> - Cutaneous neurofibromas - Lisch nodules - Spinal neurofibromas - Spine deformities (scoliosis) - Vitamin D 25-OH deficiency - Optic glioma - Astrocytoma 	G4
NF_76	M	33	<ul style="list-style-type: none"> - CALMs - Cutaneous neurofibromas - Axillary freckling - Lisch nodules - Anxiety disorder - Optic glioma 	G4
NF_81	F	30	<ul style="list-style-type: none"> - CALMs - Spinal neurofibromas - Lisch nodules - Spine deformities (scoliosis) - Anxiety and depressive disorder - Epilepsy syndrome - Hearing loss - Optic glioma - Gangliocytoma - Learning disabilities - UBOs 	G4
NF_82	F	30	<ul style="list-style-type: none"> - CALMs - Subcutaneous neurofibromas - Spinal neurofibromas - Spine deformities (scoliosis) - 25-OH Vitamin D deficiency - UBOs - Retroperitoneal ganglioneuroma 	G4
NF_85	F	24	<ul style="list-style-type: none"> - Plexiform neurofibroma along the third branches of the trigeminal nerve - Spine deformities (kyphoscoliosis) - Optic glioma 	G4
NF_99	F	47	<ul style="list-style-type: none"> - CALMs - Cutaneous neurofibromas - Anxiety disorder - Spine deformities (scoliosis) - Retroperitoneal ganglioneuroma 	G4

NF_115	F	28	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Lisch nodules - Bone deformities (foot deformities) - Optic glioma 	G4
NF_15	M	31	<ul style="list-style-type: none"> - CALMs - Cutaneous neurofibromas - Spine deformities (loss of the normal cervical lordosis) - Cardiovascular disorder (hypertension) - Retinopathy - Infundibular/hypothalamus lipoma 	G5
NF_19	F	25	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Axillary freckling - Spine deformities (loss of the normal cervical lordosis) - Depressive disorder - Pheochromocytoma/paraganglioma 	G5
NF_22	F	46	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Lisch nodules - Spine (scoliosis) and bone (osteoporosis) deformities - Cardiovascular disorders (hypertension) - Tethered Cord syndrome - Spinal dysraphism - Breast cancer 	G5
NF_23	F	57	<ul style="list-style-type: none"> - CALMs - Cutaneous neurofibromas - Bone deformities (short stature) - Breast cancer 	G5
NF_29	M	52	<ul style="list-style-type: none"> - Spinal neurofibromas - Lisch nodules - Bone deformities (foot deformities) - Epilepsy syndrome - Seminoma 	G5
NF_31	M	21	<ul style="list-style-type: none"> - CALMs - Spinal neurofibromas - Bilateral paravertebral plexiform neurofibroma - Spine deformities (scoliosis) - Brugada syndrome - Learning disabilities 	G5

			- Gallbladder adenomyomatosis	
NF_32	M	21	- CALMs - Spinal neurofibromas - Spine deformities (scoliosis) - Brugada syndrome - Adenomyomatosis	G5
NF_39	M	27	- Spine deformities (loss of the normal cervical lordosis) - Anxiety disorder - Vascular anomalies - Femur ossifying fibromas	G5
NF_42	F	44	- CALMs - Axillary and inguinal freckling - Spine (joint spondylosis) and bone (cervical spondylosis) deformities - Anxiety and depressive disorder - Sleep disorder - Nabothian cysts - Fibromioma	G5
NF_55	F	52	- CALMs - Cutaneous and subcutaneous neurofibromas - Spine (scoliosis) and bone (osteopenia and foot deformities) deformities - Hypercholesterolemia - 25-OH Vitamin D deficiency - Uterine myomas	G5
NF_73	M	37	- CALMs - Cutaneous and subcutaneous neurofibromas - Spinal neurofibroma - Lisch nodules - Spine (loss of the normal cervical lordosis) and bone (heterometry of the lower limbs) deformities - Learning disabilities - Pheochromocytoma	G5
NF_78	F	47	- CALMs - Cutaneous neurofibromas - Spine deformities (scoliosis) - Hearing loss - Uterine myoma - Cholesteatoma (left ear)	G5

NF_79	F	45	<ul style="list-style-type: none"> - Cutaneous neurofibromas - Lisch nodules - Spine deformities (scoliosis) - Dorso-lumbar CSF cyst - Tubular adenoma 	G5
NF_87	M	51	<ul style="list-style-type: none"> - Cutaneous neurofibromas - Cardiovascular disorder (hypertension) - Hypercholesterolemia - Prostate cancer 	G5
NF_92	M	56	<ul style="list-style-type: none"> - CALMs - Spinal neurofibromas - Malar facial plexiform neurofibroma - Spine deformities (lost of the normal cervical lordosis) - Cardiovascular disorders (mitral and aortic insufficiency) - Hypercholesterolemia - Lymphoma - GIST 	G5
NF_97	F	39	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Spine (scoliosis) and bone deformities (pes cavus) - Breast cancer 	G5
NF_98	M	60	<ul style="list-style-type: none"> - CALMs - Cutaneous and subcutaneous neurofibromas - Spine deformities (joint spondylosis) - Pheochromocytoma 	G5
NF_101	M	21	<ul style="list-style-type: none"> - Cutaneous neurofibromas - Spine (scoliosis) and bone (non-ossifying fibromas of tibia) deformities - Cardiovascular disorders (aortic stenosis) - Moyamoya syndrome 	G5
NF_105	M	40	<ul style="list-style-type: none"> - CALMs - Cardiovascular disorder (hypertension) - Spinal neurofibromas - Bladder heteroplasia - Left thigh and leg malignant plexiform neurofibromas 	G5

Table S1. Demographics and phenotypic features according to clinical groups (G1-G5) in the NF1 cohort. In column ID all NF1 patients were indicated with specific code (NF_ followed by an Arabic number) corresponding to the 'unique patient identifier' of the clinical centre. Age is referred to age at diagnosis. Group 1 (G1) includes patients with classical NF1 phenotype, including 6 or more café-au-lait spots (CALMs), axillary and inguinal freckling, two or more Lisch nodules and neurofibromas, without other manifestations of extra-cutaneous/ocular involvement; Group 2 (G2) encloses NF1 patients presenting the phenotypic features of G1 plus involvement of skeletal apparatus, central nervous system (epilepsy, intracranial vascular malformations, hamartomas/UBOs) and mental system (intellectual disability, anxiety/depression/sleep disorders), vascular system and anomalies of internal organs; Group 3 (G3) includes NF1 patients presenting the phenotypic features of G1 plus multi-apparatus involvement and histological diagnosis of MPNST; Group 4 (G4) includes NF1 patients presenting the phenotypic features of group G1 plus multi-apparatus involvement and neoplasms of the central and peripheral nervous systems; Group 5 (G5) includes NF1 patients presenting the clinical traits of G1 plus multi-apparatus involvement and neoplasms of variable grade of various organs and apparatus. UBOs: Unidentified bright objects; MGUS: Monoclonal Gammopathy of Undetermined Significance; MPNST: Malignant Peripheral Nerve Sheath Tumor, GIST: Gastrointestinal stromal tumor. F: female, M: male.

ID	NF1 genetic mutation	Technique			NGS			
		NGS	RT-PCR/Sanger sequencing	MLPA	Depth of the identified variants (TOT:WT:MUT)	Average depth	Depth minimum	Depth maximum
NF_1	c.(586+1_587-1)_(730+1_731-1)del	✓				125,15	15	297
NF_2	c.3G>A, p.?	✓			11:3:8	146,39	10	413
NF_3	c.3G>A, p.?		✓					
NF_4	c.3G>A, p.?		✓					
NF_5	c.1595T>G, p.Leu532Arg		✓					
NF_6	c.479+5G>A , p.Leu94fs	✓			91:47:44	132,61	15	340
NF_8	c.7884_7885del, p.(Phe2629Serfs*9)	✓			79:33:46	106,34	20	299
NF_10	c.3826C>T, p.Arg1276*		✓					
NF_11	c.3826C>T, p.Arg1276*		✓					
NF_13	c.6892_6897del, p.Ala2298_Val2299del	✓			249:142:107	217,75	13	651
NF_15	c.2619dup, p.Lys874*	✓			42:20:22	144,69	22	394
NF_17	c.6364+4A>G , p.Val2029Lysfs*7		✓					
NF_18	c.3326T>G, p.Leu1109*		✓					
NF_19	c.3826C>T , p.Arg1276*		✓					
NF_20	c.1783_1784del, p.Glu595fs		✓					
NF_21	c.(?-383)_(*3522_?)del (whole NF1 deletion)	✓				99.5	15	185
NF_22	c.4982_4983del, p.Cys1661*	✓			123:68:55	129,83	26	373
NF_23	c.3496+1G>A , p.Tyr1106Leufs*28		✓					
NF_25	c.3496+1G>A, p.Tyr1106Leufs*28		✓					
NF_26	c.1246C>T, p.R416*		✓					

NF_28	c.1A>G, p.?	✓			13:1:12	132,34	15	382
NF_29	c.1595T>G, p.Leu532Arg		✓					
NF_30	c.3728T>C, p.Leu1243Pro	✓			252:135:117	121,39	17	343
NF_31	c.7686delG, p.Ile2563Phefs*40		✓					
NF_32	c.7686delG, p.Ile2563Phefs*40		✓					
NF_35	c.4269+2T>C, p.?		✓					
NF_36	c.6335T>C, p.Leu2112Pro		✓					
NF_37	c.(?_-383)_(*3522_?)del (whole NF1 deletion)	✓				96,21	12	175
NF_38	c.4768C>T, p.Arg1590Trp		✓					
NF_39	c.1009G>T, p.Glu337*	✓			261:130:131	172,53	22	495
NF_40	c.1466A>G, p.Tyr489Cys	✓			60:25:35	185,38	45	498
NF_41	c.1499_1501delinsAAA, p.Ile500_His501delinsLysAsn		✓					
NF_42	c.3502-3519 del, p.G1169-L1173del		✓					
NF_43	c.2352G>C, p.Trp784Cys	✓			26:11:15	122,02	52	352
NF_45	c.2307dup, p.Thr770HisfsTer6	✓			254:132:122	152,66	10	401
NF_46	c.2307dup, p.Thr770HisfsTer6	✓			254:132:122	150,09	11	419
NF_47	c.2307dup, p.Thr770HisfsTer6	✓			190:106:84	149,78	17	422
NF_48	c.1A>G, p.?	✓			23:9:14	142,97	24	391
NF_50	c.3497_3974del		✓					
NF_51	c.4381dup, p.Ile1461Asnfs*4	✓			63:35:28	124,44	25	335
NF_52	c.4309G>T, p.(Glu1437*)	✓			60:26:34	122,34	12	442
NF_53	c.1378dup, p.Ile460Asnfs*10	✓			212:108:104	156,18	11	426
NF_54	c.1639G>T, p.Glu547*	✓			81:38:43	162,43	10	442
NF_55	c.4537C>T, p.Arg1513*	✓			49:23:26	181,98	33	495
NF_56	c.2409+1G>A, p.?.; c.2375T>A, p.Leu792His	✓			33:17:16	158,46	32	445
NF_57	c.1381C>T, p.Arg461*	✓			167:85:82	134,68	21	374
NF_59	c.4270-2A>G, p.Ile1424_Gln1426del	✓			46:28:18	144,01	20	402

NF_60	c.1756_1759del, p.Thr586Valfs*18	✓			35:16:19	125,90	19	339
NF_61	c.4367+2T>C , p.?		✓					
NF_62	c.1260+1G>A, p.Ser421fs	✓			101:58:43	172,98	17	488
NF_63	c.4733C>A, p.(Ser1578Tyr)	✓			159:79:80	194,49	16	509
NF_65	c.(?-383)_(*3522_?)del (whole NF1 deletion)	✓				82,03	13	154
NF_66	c.2665 A>G, p.Thr889Ala	✓			54:35:19	168,88	15	415
NF_67	c.6084+1G>A , p.?	✓			199:105:94	224,34	26	542
NF_68	c. 6792C>A , p.Ala2253_Lys2286del		✓					
NF_70	c.2409+1G>C , p.?	✓			35:24:11	196,06	32	510
NF_71	c.2409+1G>C , p.?	✓			53:28:25	184,97	25	506
NF_72	c.7884_7885del, p.Phe2629Serfs*9	✓			242:126:116	237,30	11	592
NF_73	c.4923G>A, p.Trp1641*		✓					
NF_76	c.1845+1_1845+5del, p.Ala548_Lys615del	✓			92:47:45	194,93	15	529
NF_77	c.2326G>A, p.Ala776_Gln803del		✓					
NF_78	c.2329T>C, p.Trp777Arg	✓			37:23:14	226,59	14	592
NF_79	c.3916C>T, p.Arg1306*, c.1975C>T, p.Arg659Trp	✓			145:71:74	211,39	10	544
NF_80	c.4278G>C, p. Gln1426His	✓			58:30:28	151,59	15	385
NF_81	c.5819del, p.(Lys1940Serfs*18)	✓			131:61:70	175,05	19	459
NF_82	c.8051-1 G>C , p.?	✓			63:23:40	142,41	17	387
NF_83	c.5673T>G, p.Ser1891Arg	✓			111:60:51	221,46	13	587
NF_85	c.6621dup, p.Trp2208Valfs*13	✓			285:145:140	162,50	12	423
NF_86	c.4923G>A, p.Trp1641*	✓			156:78:78	181,44	11	431
NF_87	c.7259C>A, p.Ala2420Asp		✓					
NF_89	c.(4661+1_4662-1)_(7258+1_7259-1)dup	✓		✓				
NF_90	c.(4661+1_4662-1)_(7258+1_7259-1)dup	✓		✓				
NF_92	c.4278G>C. p.Gln1426His	✓			71:35:36	165,75	26	447
NF_94	c.2252-3T>G, p.?	✓			195:85:110	134,13	24	369

NF_96	c.(?-383)_(*3522_?)del (whole <i>NF1</i> deletion)	✓				79,50	12	165
NF_97	c.4515-2A>G , p.?	✓			28:5:23	151,35	22	395
NF_98	c.7089dup, p.Asn2364*		✓					
NF_99	c.2851G>T, p.Val951Phe		✓					
NF_100	c.4537C>T , p.Arg1513*	✓			41:50:11	142,88	21	394
NF_101	c.1329delT, p.Phe443Leufs*29		✓					
NF_102	c.1381C>T, p.Arg461*		✓					
NF_103	c.2251 G>C , p.Asp668Glufs*9		✓					
NF_105	c.C7532T, p.(Ala2532Val)		✓					
NF_115	c.2446C>T, p.Arg816*		✓					
NF_127	c.2540T>G, p.Leu847Arg		✓					

Table S2. Sequencing techniques performed and NGS details. Next-generation sequencing (NGS), Real Time PCR (RT-PCR), Sanger sequencing, and Multiple Ligation Probe Amplification (MLPA) were applied to *NF1* mutation analysis. NGS details including depth of the identified *NF1* variants, average depth, minimum and maximum depth are shown.