

Supplementary Table S1. Relatives with NF of Spanish participants

	n	%
No relatives with NF	264	77.65
Brother/sister with NF^a	30	8.82
<i>Number of affected siblings</i>		
<i>One</i>	23	6.76
<i>Two</i>	5	1.47
<i>Three</i>	1	0.29
<i>MD</i>	1	0.29
Son/daughter with NF^b	28	8.24
<i>Mean of children with NF</i>		
<i>One</i>	26	7.65
<i>Two</i>	2	0.59
Father with NF^c	25	7.35
Mother with NF^d	22	6.47
Other relatives with NF	30	8.82

NF: Neurofibromatosis; MD: missing data.

a) NF1=29; SWN=1; b) all NF1; c) NF1=23, NF2=1; SWN=1; d) all NF1

Supplementary Table S2. Patients' emotions and feelings concerning NF

Feeling/Emotion	Spain		Portugal	
	n	%	n	%
Acceptance	121	35.59	22	37.93
Uncertainty	98	28.82	13	22.41
Resignation	94	27.65	3	5.17
Fear or concern	89	26.18	26	44.83
Impotence	46	13.53	9	15.52
Isolation or loneliness	44	12.94	6	10.34
Frustration	44	12.94	14	24.14
Sadness, grief	44	12.94	3	5.17
Incomprehension	36	10.59	10	17.24
Depression	32	9.41	2	3.45
Pain	28	8.24	5	8.62
Hope	27	7.94	7	12.07
Shame	27	7.94	1	1.72
Anger	25	7.35	1	1.72
Indifference	22	6.47	1	1.72
Don't know/No answer	16	4.71	7	12.07
Desperation	15	4.41	2	3.45
Nuisance	12	3.53	2	3.45

Bold means: Main emotions concerning NF patients. Highlighted means: those over 20% in each country

Supplementary Table S3. Work and educational impact of neurofibromatosis on Spanish and Portuguese participants

Work/academic impact and dependency	Spain			Portugal		
	n	%	N	n	%	N
Difficulties in access to work due to NF (>16 years)	57	30.48	187	9	26.47	34

Difficulties in continuing studies due to NF (>16 years)	93	39.91	233	12	35.29	34
Need for social worker support	88	25.88	340	14	24.14	58
Patients who have received care from social workers	68	77.27	88	10	71.43	14
Company awareness of the disease						
<i>Yes</i>	49	36.57	134	16	69.57	23
<i>No</i>	27	20.15	134	3	13.04	23
<i>DK/DA</i>	58	43.28	134	4	17.39	23

Supplementary Table S4. Caregivers' socio-demographic characteristics

Variable	Spain		Portugal	
	n	%	n	%
Caregiver's gender				
<i>Female</i>	114	74.03	21	84.00
<i>Male</i>	40	25.97	4	16.00
Caregiver's relationship				
<i>Father/Mother</i>	150	96.15	25	100.00
<i>Partner</i>	3	1.92	0	0.00
<i>Brother/sister</i>	1	0.64	0	0.00
<i>Uncle/Aunt</i>	1	0.64	0	0.00
<i>Other</i>	1	0.64	0	0.00
<i>Grandfather/Grandmother</i>	0	0.00	0	0.00
<i>Not relative</i>	0	0.00	0	0.00
Patients in charge				
<i>One patient</i>	147	94.23	25	100.00
<i>Two patients</i>	7	4.49	0	0.00
<i>Three patients</i>	1	0.64	0	0.00
<i>Missing data</i>	1	0.64		

Supplementary Table S5. Impact of NF on caregivers

Caregiver's impairment	Never		Almost never		Occasionally		Almost always		Always	
	n	%	n	%	n	%	n	%	n	%
"I feel that due to the time I dedicate to the NF patient, I no longer have enough time for myself."										
Spain	67	43.51	19	12.34	41	26.62	22	14.29	5	3.25
Portugal	5	20.00	6	24.00	11	44.00	3	12.00	0	0.00
"I feel that due to the time required by the NF sufferer I do not dedicate enough time to the rest of the family."										
Spain	66	42.86	25	16.23	44	28.57	17	11.04	2	1.30
Portugal	7	28.00	8	32.00	8	32.00	2	8.00	0	0.00
"I feel very overburdened by the care required by the person in my care in relation to NF."										
Spain	67	43.51	30	19.48	38	24.68	15	9.74	4	2.60
Portugal	7	28.00	6	24.00	11	44.00	1	4.00	0	0.00
"I feel that, due to NF, family activities have been limited (e.g., family vacations, outings, celebrations, extracurricular activities, etc.)."										
Spain	77	50.00	24	15.58	30	19.48	15	9.74	8	5.19
Portugal	11	44.00	8	32.00	5	20.00	1	4.00	0	0.00

Supplementary Table S6. Caregivers' work impairment concerning NF

Type of work impairment	Spain		Portugal	
	n	%	n	%
Days of leave	31	47.69	4	50.00
Reduction of working hours	20	30.77	5	62.50
Abandonment or loss of employment	17	26.15	1	12.50
Leave of absence	5	7.69	0	0.00
Early retirement	31	47.69	0	0.00

Supplementary Table S7. Frequency of medical visits

Variable	Spain			Portugal		
	Mean	SD	n	Mean	SD	n
Frequency of visits (months)	9.01	6.65	313	6.44	5.40	54
NF1	9.14	6.77	288	6.80	5.41	50
NF1 <18 years old	6.82	3.80	109	6.42	6.89	24
NF1 >18 years old	10.18	7.51	204	7.15	3.67	26
NF2	7.27	5.15	22	2.00	2.71	4
SWN	9.33	3.06	3	2.33	1.76	
Number of visits since diagnosis	3.77	4.29	340	2.43	1.79	58
NF1	3.76	4.41	314	1.00	0.00	54
NF2	3.74	2.60	23			4
SWN	4.33	1.15	3			

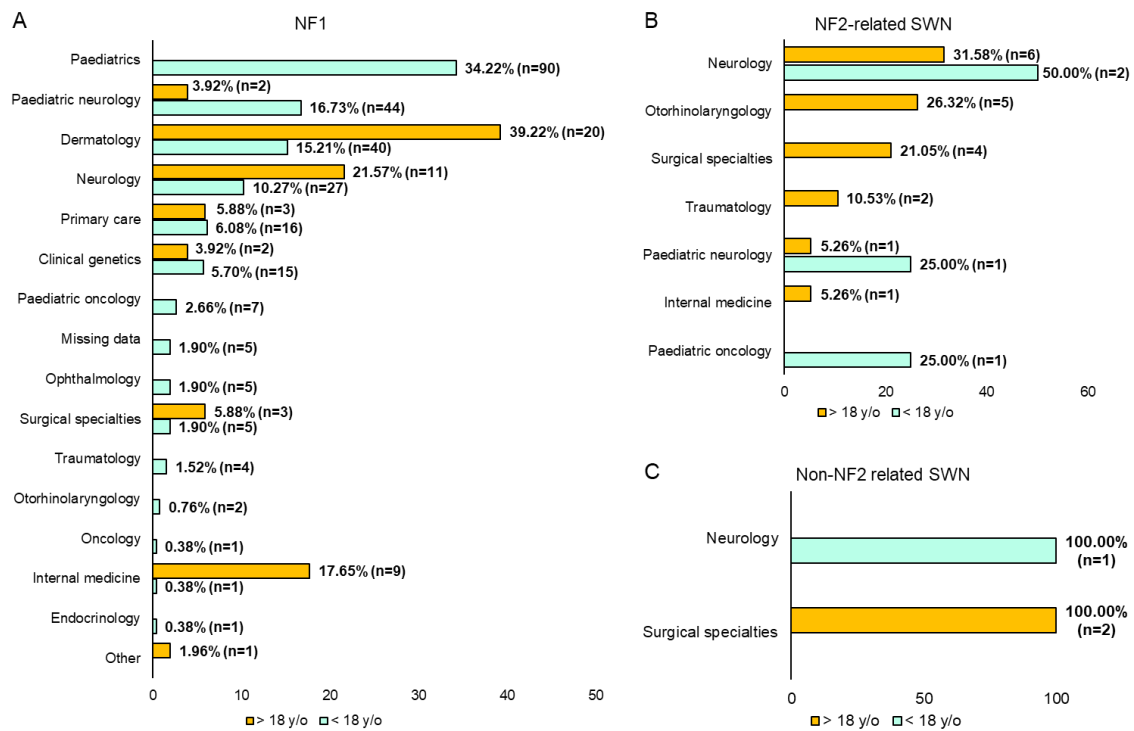
Supplementary Table S8. Clinical management of NF

Pathology management	Spain			Portugal		
	n	%	N	n	%	N
Healthcare setting						
Public healthcare	299	87.91	340	49	84.48	340
Hospital	282	94.31	299	47	95.92	49
Health centre	17	5.68	299	2	4.08	49
Occasional recourse to private healthcare	116	38.80	299	0	0.00	49
Private health care	26	7.65	340	5	8.62	340
None	15	4.41	340	4	6.90	340
Treatment						
Any therapy from symptom onset	118	34.71	340	25	43.10	58
Pharmacological treatment currently	87	25.59	340	20	34.48	58

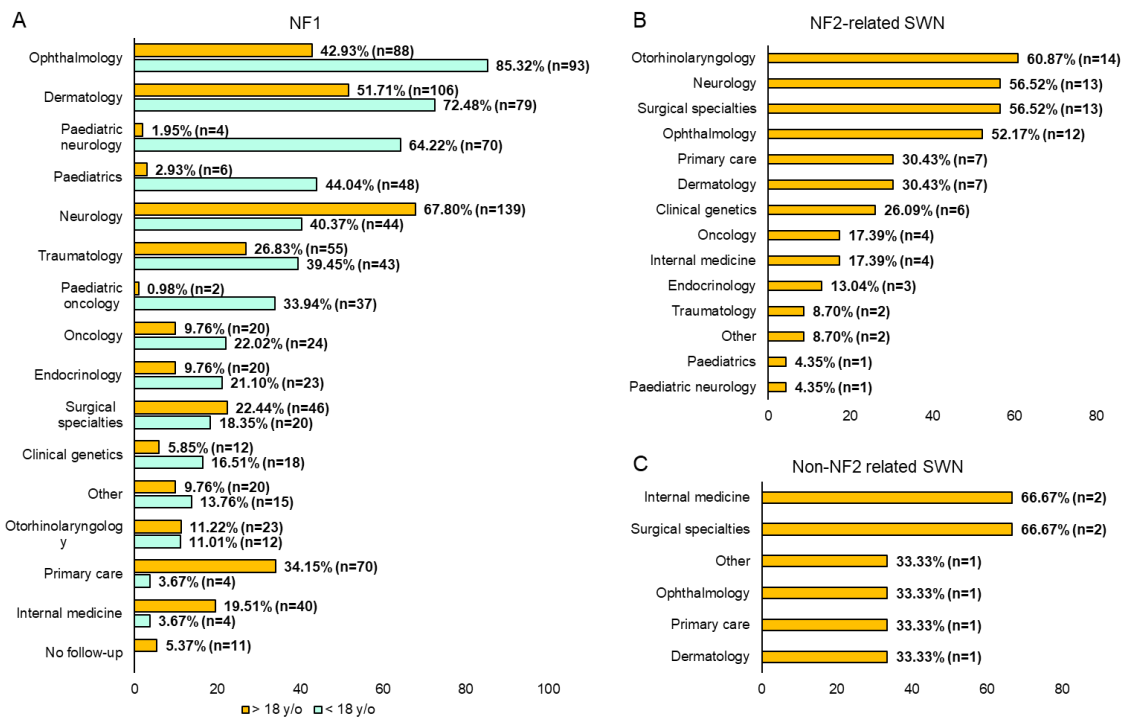
Pharmacological treatment for PN currently	24	17.65	136	6	40.00	15
NF-related surgeries	214	62.94	340	25	43.10	58
PN-related surgeries	82	60.29	136	4	66.67	6
Hospitalization in the last 12 months	40	11.76	340	7	12.07	58

NF: Neurofibromatosis ; PN : plexiform neurofibroma

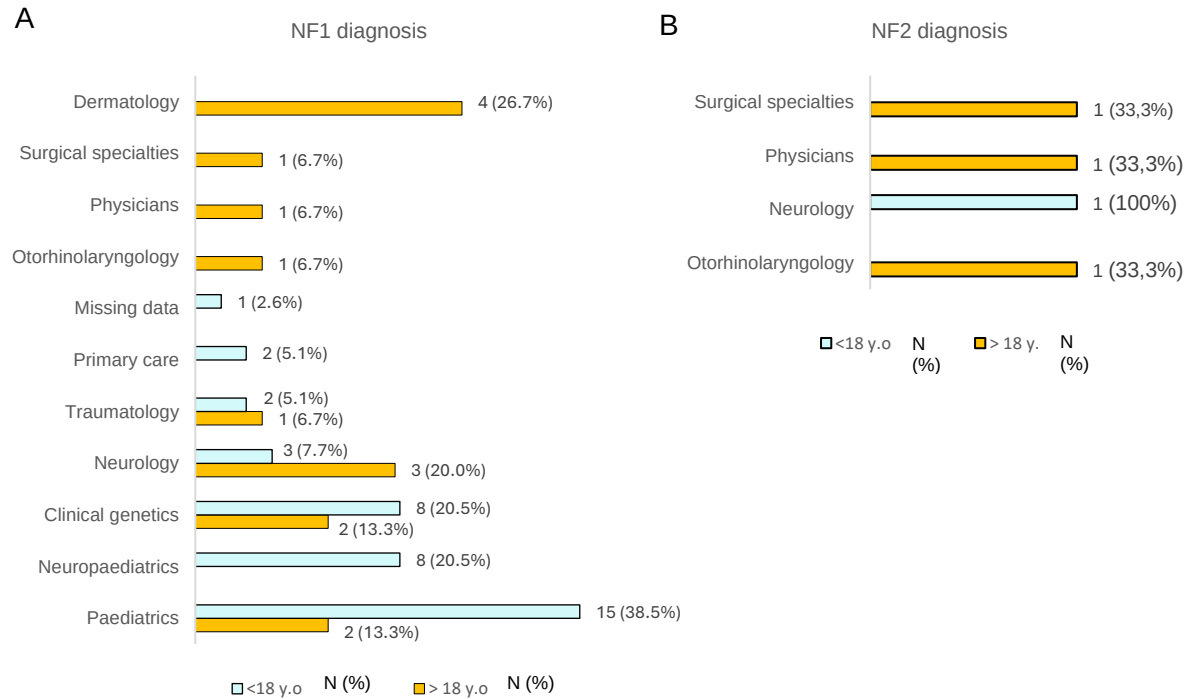
Supplementary Figure 1. Medical specialties involved in the diagnosis of Spanish patients with NF1 (A), NF2-related (B) and non-NF2 related SWN (C). NF: neurofibromatosis; SWN: schwannomatosis.



Supplementary Figure 2. Medical specialities involved during the follow-up of Spanish patients with NF1 (A), NF2-related (B) and non-NF2 related SWN (C). NF: neurofibromatosis; SWN: schwannomatosis.

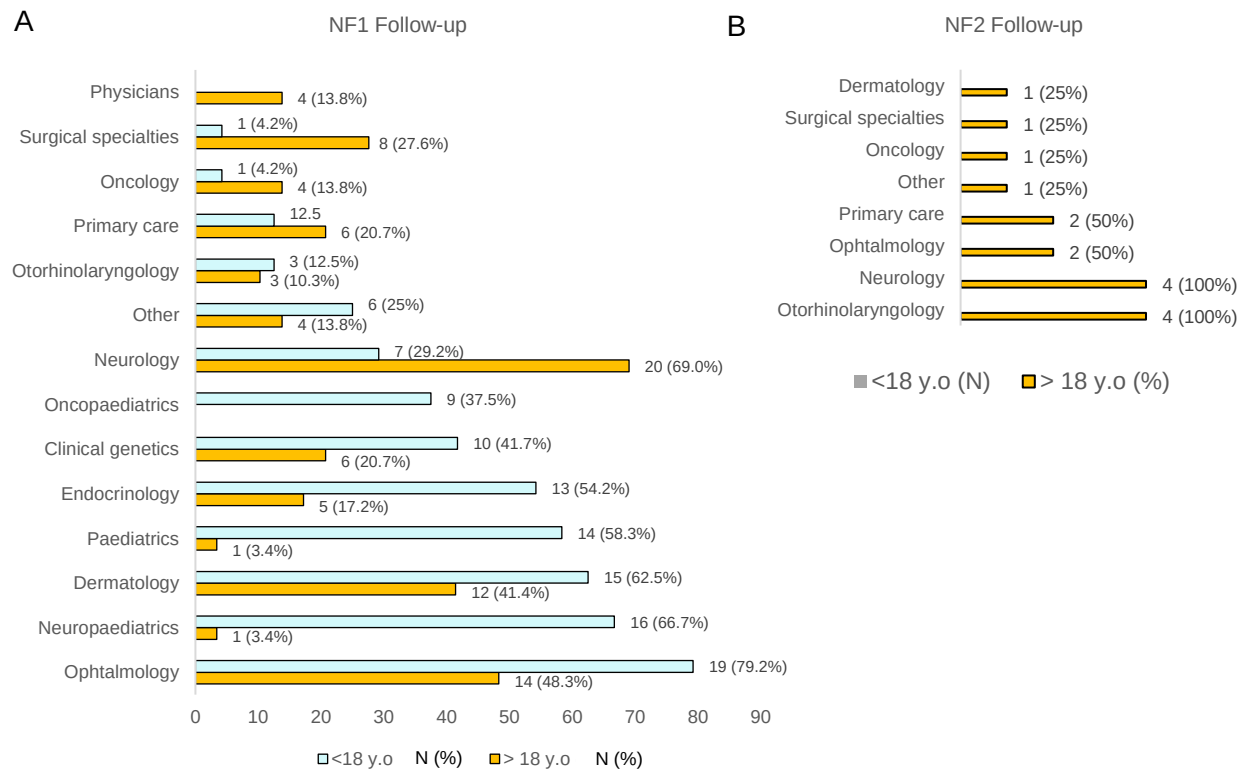


Supplementary Figure 3. Medical specialties involved in diagnosing Portuguese patients with NF1 (A) and NF2-related SWN (B). NF: neurofibromatosis; SWN: schwannomatosis.



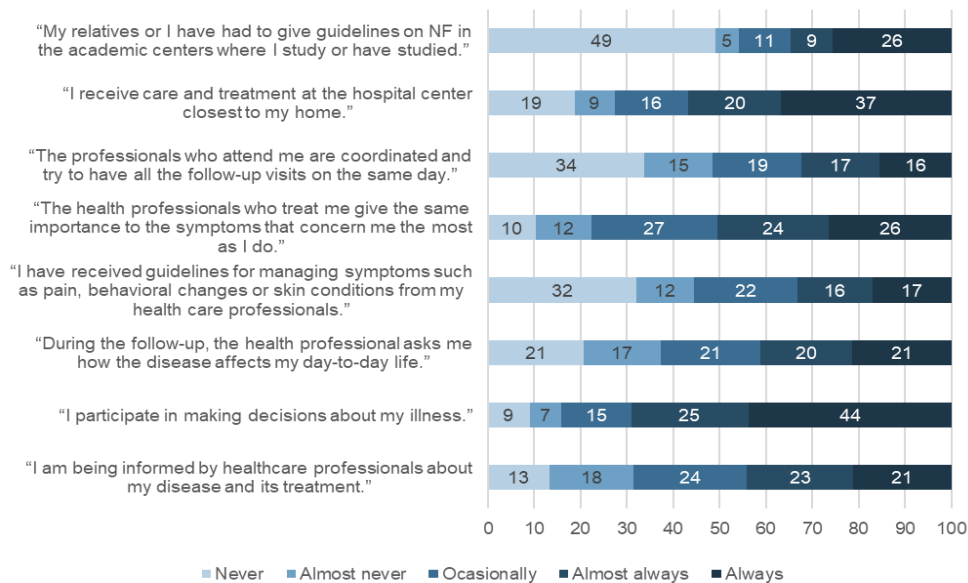
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Supplementary Figure 4. Medical specialties involved during the follow-up of Portuguese patients with NF1 (A) and NF2-related SWN (B). NF: neurofibromatosis; SWN: schwannomatosis



Supplementary Figure 5. The frequency (%) with which Spanish (A) and Portuguese (B) participants have identified different issues related to the management of NF.

A



B

