SUPPLEMENTARY MATERIAL

Whole-genome sequencing in Brazilian patients with neurofibromatosis type 1, including novel variants, incidental findings, and dual diagnoses

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Table 1S. Sex, age at first appointment for genetic evaluation, family history, and clinical findings of the 50 individuals with NF1 included in the present study

Family/patient		FH	Sex	Age	CALMs	LMs	cNF	pNF	LNs	OPG	SWD	CPT	Other
1	Proband	-	M	1y	+	+	-	-	-	-	-	-	ADHD
2	Proband	+	F	1y11m	+	+	-	-	+	-	-	-	
	Brother		M	10y	+	+	-	+	+	-	-	-	Mild ID; 47,XXY/45,X[44:6]; pathogenic variant in *TMEM127*
	Mother		F	32y	+	+	+	-	-	-	-	-	Pathogenic variant in *TMEM127*
	Father		M	31y	+	+	+	+	+	-	-	-	
3	Proband	-	M	11y	+	+	+	+	+	-	-	-	Dysembryoplastic neuroepithelial tumor at 19y
4	Proband	-	F	19y	+	+	+	-	+	-	-	-	LD, mild ID, congenital hip dislocation
5	Proband	+	M	9y	+	+	+	-	+	-	-	+	Pathogenic variant in *BRCA1*
	Mother		F	30y	+	+	+	+	+	-	-	-	Pathogenic variant in *BRCA1*, cholangiocarcinoma at 33y and 47y; died at 51y
6	Proband	?	F	40y	+	+	+	-	?	-	-	-	
7	Proband	+	F	30y	+	+	+	+	+	-	-	-	
	Son		M	12y	+	+	+	-	+	-	-	-	
	Daughter		F	4y	+	+	-	+	+	+	-	-	
8	Proband	-	F	2y	+	+	-	-	+	-	-	-	
9	Proband	-	F	47y	+	+	+	+	+	-	-	-	Breast cancer (unilateral invasive ductal carcinoma) at 51y Cerebellar ataxia at 47y (variant in *KCND3*).
10	Proband	+	F	24y	+	+	+	-	+	-	-	-	
	Mother		F	46y	+	+	+	-	+	-	-	-	MPNST of the right thigh at 45y
	Uncle		M	62y	+	+	+	-	+	-	-	-	
11	Proband	+	M	18y	+	+	+	-	+	-	-	-	
	Mother		F	43y	+	+	+	+	+	-	-	-	Basal cell carcinoma at 43y
	Brother		M	24y	+	+	+	-	+	-	-	-	
12	Proband	-	M	13y	+	+	+	+	-	-	-	-	
13	Proband	-	F	8y	+	+	+	+	+	-	-	-	
14	Proband	+	M	12y	+	+	-	-	-	-	-	-	
	Mother		F	48y	+	+	-	-	?	-	-	-	
	Sister 1		F	22y	+	+	+	-	?	-	-	-	
	Sister 2		F	20y	+	+	-	-	?	-	-	-	
	Brother		M	17y	+	+	-	-	?	-	-	-	

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Family/patient		FH	Sex	Age	CALMs	LMs	cNF	pNF	LNs	OPG	SWD	CPT	Other
15	Proband	-	M	9m	+	+	-	-	+	-	-	-	
16	Proband	-	F	31y	+	+	+	+	-	-	-	-	Breast cancer at the age of 38y
17	Proband	-	F	21y	+	+	+	+	+	-	-	-	
18	Proband	-	M	50y	+	+	+	+	+	-	-	-	Mild ID, MPNST of the dorsum at 51y, died at 53y
19	Proband	+	F	2y	+	+	-	-	+	-	-	-	
	Uncle		M	18y	+	+	+	+	+	-	-	-	
	Cousin		F	1y	+	-	-	-	-	-	-	-	
20	Proband	-	M	15y	+	+	-	-	-	-	-	-	Neurofibromatosis-Noonan phenotype
21	Proband	-	F	16y	+	+	-	-	-	-	-	-	
22	Proband	-	F	4y	+	+	+	-	+	-	-	-	
23	Proband	-	F	4y	+	+	+	+	+	-	-	-	
24	Proband	-	M	37y	+	+	+	+	+	-	-	-	
25	Proband	+	M	15y	+	+	+	-	-	-	-	-	
	Mother		F	51y	+	+	+	+	+	+	+	-	
26	Proband	+	F	30y	+	+	+	-	+	-	-	-	Pilocytic astrocytoma of the left posterior fossa at 29y
	Mother	+	F	59y	+	+	+	+	+	-	-	-	
27	Proband	+	M	2y	+	-	-	-	-	-	-	-	DD, macrocrania
	Mother		F	32y	+	+	-	-	+	-	-	-	LD, macrocrania
	Sister		F	5y	+	-	-	-	?	-	-	-	
28	Proband	-	F	29y	+	+	+	+	-	+	-	-	
29	Proband	+	F	3y10m	+	+	-	-	-	-	-	+	DD
30	Proband	-	M	11m	+	+	-	-	-	-	-	-	*Situs inversus totalis*, recurrent pulmonary infections (variants in *DNAH5*)

+: present; -: absent; ?: uncertain or unknown; ADHD: attention deficit hyperactivity disorder; cNF: cutaneous neurofibromas; CPT: congenital pseudoarthrosis of the tibia; DD: developmental delay; F: female; FH: Family history; CALM: café-au-lait macules; ID: intellectual deficiency; LD: learning disability; LMs: lentiginous macules; LNs: Lisch nodules; m: months; M: male; MPNST: malignant peripheral nerve sheath tumor; OPG: optic pathway glioma; pNF: plexiform neurofibroma(s); SWD: sphenoid wing dysplasia; y: years.