

Barriers to disclosure of diagnoses and diagnostic testing in families affected by X-linked adrenoleukodystrophy (Schäfer, L.; Deibler, L.; Lier, J.; Ponleitner, M.; Köhler, W.; Bergner, C-C.)

Table S1: Characteristics of participants and pedigree family members, including frequencies of not informed and not tested potentially affected family members (PAFMs).

Characteristics of the participant										Description of the participant's pedigree chart regarding the phenotype frequency of ALD-positive family members (including participant)												Frequency of not informed or informed but not tested PAFMs in the pedigree chart (excluding index patient)				Description of not informed and/or not tested PAFMs regarding sex and age			
Case No.	Sex	Age in years	Years since diagnosis	Relationship to index patient	Phenotype	HSCT	AACS	EDSS	Genotype	No. of ALD-positive FM	No. of boys with CCALD	No. of boys deceased from CCALD	No. of asymptomatic boys	No. of men with ACALD + AMN	No. of men deceased from ACALD	No. of men with AMN	No. of asymptomatic men	No. of asymptomatic girls	No. of women with myeloneuropathy	No. of asymptomatic women	No. of FM with unknown symptom status	No. of PAFM	No. of tested PAFM	No. of not informed PAFM	No. of informed, but not tested PAFM	No. of boys (<18 y)	No. of men (≥18 y)	No. of girls (<18 y)	No. of women (≥18 y)
1	male	66	23	brother	AMN	no	8.0	6.5	c.1876G>A (p.Ala626Thr)	8	0	0	0	0	0	3	0	2	1	0	2	22	7	13	2	0	3	2	10
2	female	39	10	mother	asymptomatic	no	n/a	n/a	c.1632G>T (p.Gin544His)	2	0	0	0	0	0	0	0	1	1	0	0	14	1	8	5	0	5	1	7
3	female	64	32	sister	myeloneuropathy	no	3.0	n/a	n/a	6	1	1	0	1	1	1	0	0	1	2	0	7	5	2	0	0	1	0	1
4	female	46	20	sister	myeloneuropathy	no	3.5	n/a	c.1905del5; n/a	3	0	0	0	0	0	1	0	0	2	0	0	8	2	5	1	0	2	0	4
5	male	47	23	self	ACALD + AMN	no	6.0	6.0	c.428C>T (p.Pro143Leu)	9	2	1	0	1	0	2	0	2	1	1	0	11	4	4	2	1	2	1	2
6	male	32	5	self	AMN	no	7.0	6.0	c.385dup (p.Ala129Glyfs*66)	1	0	0	0	0	0	1	0	0	0	0	0	8	7	0	1	0	0	0	1
7	male	56	29	brother	AMN	no	7.0	6.5	c.(1082+1_1083-1)_(1488+1_1489_1)del (p.Asp361Glyfs*59)	9	1	1	1	0	0	2	0	0	2	3	0	11	11	0	0	0	0	0	0
8	male	52	17	self	ACALD+ AMN	yes	9.0	5.5	n/a	1	0	0	0	1	0	0	0	0	0	0	0	6	0	5	1	0	1	0	5
9 ^a	female	36	24	daughter	asymptomatic	no	0.0	0.0	c.[524_526delTCT;901_2235dup] (p.[Phe175del;Val301_Thr745dup])	6	0	0	0	3	1	0	0	0	2	1	0	8	7	0	1	0	0	0	1
10	female	44	19	daughter	myeloneuropathy	no	1.5	1.5	c.799G>A (p.Clu267Lys)	4	0	0	0	0	0	2	0	0	2	0	0	12	7	0	5	0	0	5	0
11 ^a	male	49	24	nephew	ACALD+ AMN	yes	n/a	n/a	c.[524_526delTCT;901_2235dup] (p.[Phe175del;Val301_Thr745dup])	6	0	0	0	3	1	0	0	0	2	1	0	9	9	0	0	0	0	0	0
12	female	46	5	mother	myeloneuropathy	no	2.0	n/a	c.1978C>T (p.Arg660Trp)	5	0	0	0	0	0	1	0	0	3	1	0	12	5	0	7	0	2	0	5
13 ^b	male	45	13	first cousin	Morbus Addison only	no	1.0	1.0	c.454C>T (p.Arg152Cys)	10	1	1	0	0	0	3	1	0	1	4	0	10	9	0	1	0	1	0	0
14	female	68	33	third cousin	myeloneuropathy	no	n/a	n/a	c.1522C>T (p.Pro508Ser)	6	1	1	0	0	0	1	0	0	4	0	0	9	9	0	0	0	0	0	0
15	female	35	13	daughter	myeloneuropathy	no	n/a	n/a	n/a	9	0	0	0	0	0	4	0	0	2	3	0	17	14	0	3	0	3	0	0
16	female	61	23	mother	myeloneuropathy	no	3.0	3.0	n/a	3	1	0	0	0	0	0	0	0	2	0	0	14	12	0	2	0	1	0	1
17	female	65	13	second cousin	myeloneuropathy	no	9.0	n/a	n/a	23	1	1	1	0	0	7	0	1	10	0	2	38	32	0	6	1	1	0	4
18	female	63	13	mother	myeloneuropathy	no	5.0	3.5	c.1780G>C (p.Gly594Arg)	4	1	0	0	0	0	0	0	0	2	1	0	4	2	0	2	0	0	0	2
19	male	66	32	self	AMN	no	13.0	7.5	c.1451C>T (p.Pro484Leu)	3	0	0	0	0	0	1	0	0	1	1	0	6	2	1	3	0	1	2	1
20	female	34	7	mother	myeloneuropathy	no	n/a	n/a	n/a	2	1	1	0	0	0	0	0	0	1	0	0	7	5	1	0	0	0	0	1
21	male	74	9	self	AMN	no	8.0	6.5	c.1522C>T (p.Pro508Ser)	1	0	0	0	0	0	1	0	0	0	0	0	2	1	0	1	0	0	0	1
22	female	54	6	mother	myeloneuropathy	no	3.0	3.5	c.681C>A (p.Tyr227*)	2	1	1	0	0	0	0	0	0	1	0	0	10	8	1	1	0	1	0	1
23 ^c	male	35	11	self	ACALD+ AMN	yes	3.0	2.0	c.1534G>A (p.Gly512Ser)	10	5	5	0	1	0	0	0	1	0	3	0	13	8	0	5	0	3	0	2
24	female	55	7	mother	myeloneuropathy	no	2.5	1.5	c.1933del (p.Gln645Argfs*46)	2	0	0	0	1	1	0	0	0	1	0	0	6	5	0	1	0	0	0	1
25	female	65	24	mother	myeloneuropathy	no	7.0	6.5	n/a	4	2	2	0	0	0	0	0	0	2	0	0	8	7	0	1	0	0	0	1
26	female	41	25	daughter	asymptomatic	no	n/a	n/a	c.314C>T (p.Thr105Ile)	9	0	0	0	0	0	5	0	0	2	2	0	18	6	11	1	3	3	2	4
27	male	42	29	second cousin	AMN	no	4.0	3.5	c.1415_1416delAG (p.Gln472Argfs*83)	10	2	1	0	0	0	2	0	1	1	4	0	12	11	0	1	0	0	0	1
28	female	46	1	niece	asymptomatic	no	n/a	n/a	c.1780G>A (p.Gly594Ser)	12	1	0	0	0	0	3	0	1	0	7	0	15	11	2	2	0	0	1	3
29 ^c	female	30	9	sister	asymptomatic	no	0.0	0.0	c.1534G>A (p.Gly512Ser)	10	5	5	0	1	0	0	0	1	0	3	0	15	6	0	9	2	3	0	4
30	female	54	15	mother	myeloneuropathy	no	8.0	6.0	c.99C>G (p.Tyr33*)	4	2	1	0	0	0	0	0	0	1	1	0	9	9	0	0	0	0	0	0
31	female	65	27	mother	myeloneuropathy	no	n/a	n/a	n/a	6	2	1	0	1	1	1	0	0	2	0	0	30	6	11	13	1	9	3	10
32	female	31	28	daughter	myeloneuropathy	no	2.0	n/a	c.829G>A (p.Gly277Arg)	2	0	0	0	1	1	0	0	0	1	0	0	5	4	0	1	0	0	1	0
33	male	37	25	brother	ACALD+ AMN	yes	10.0	6.5	c.1523C>T (p.Pro508Leu)	7	1	0	0	2	1	1	0	0	2	1	0	14	9	0	5	1	2	1	1
34	female	69	18	mother	myeloneuropathy	no	7.0	6.5	c.1678C>T (p.Pro560Ser)	7	0	0	0	1	0	1	0	0	3	1	0	11	10	0	1	0	0	1	0
35	female	41	1	mother	asymptomatic	no	n/a	n/a	c.539A>G (p.Tyr180Cys)	2	0	0	1	0	0	0	0	0	0	1	0	11	4	6	1	0	3	1	3
36 ^p	male	39	35	cousin	AMN	no	3.0	3.5	c.320T>C (p.Leu107Pro)	11	2	2	0	1	1	2	0	1	1	3	1	14	14	0	0	0	0	0	0
37	male	54	16	self	ACALD+ AMN	no	6.0	6.5	c.1780G>A (p.Gly594Ser)	1	0	0	0	1	0	0	0	0	0	0	0	10	0	3	7	0	5	0	5
38	male	53	13	self	AMN	no	10.0	7.0	c.346G>C (p.Gly116Arg)	1	0	0	0	0	0	1	0	0	0	0	0	1	0	0	1	0	0	0	1
39	female	71	6	self	myeloneuropathy	no	6.0	n/a	c.2030C>A (p.Gly677Asp)	1	0	0	0	0	0	0	0	0	1	0	0	1	0	0	1	0	1	0	0
40	female	61	20	mother	myeloneuropathy	no	8.5	n/a	n/a	4	0	0	0	0	0	1	0	0	3	0	0	14	10	0	4	0	1	0	3
41	male	35	26	grandson	ACALD+ AMN	yes	1.5	2.0	c.1523C>T (p.Pro508Leu)	12	2	1	0	2	0	1	1	1	3	2	0	16	11	0	5	0	4	1	0
42	female	66	32	mother	myeloneuropathy	no	1.5	n/a	c.1528G>A (p.Gly510Ser)	4	1	1	0	0	0	1	0	0	1	1	0	6	6	0	0	0	0	0	0
43 ^b	female	71	12	second cousin	myeloneuropathy	no	3.0	n/a	c.454C>T (p.Arg152Cys)	14	1	1	0	0	0	6	0	1	1	5	0	32	16	8	8	0	5	1	10
44 ^p	male	47	27	brother	AMN	no	0.5	1.5	c.320T>C (p.Leu107Pro)	19	5	4	1	0	0	3	1	1	2	3	3	26	18	1	7	0	1	0	7
45	female	55	27	mother	myeloneuropathy	no	2.0	n/a	n/a	10	1	0	0	0	0	2	1	0	1	5	0	14	11	0	3	0	2	0	1
46	female	70	34	sister	myeloneuropathy	no	13.0	n/a	c.355del; (p.Ala119Profs*79)	13	4	4	0	0	0	4	0	0	1	4	0	8	8	0	0	0	0	0	0
47	male	49	44	brother	AMN	no	5.0	4.0	c.1451G>G (p.Pro484Arg)	6	2	2	0	2	2	1	0	0	1	0	0	25	10	0	15	1	5	2	7
48	male	57	28	second uncle	AMN	no	7.0	6.5	c.1252C>T (p.Arg418Trp)	7	0	0	0	0	0	3	0	0	1	3	0	16	9	0	7	0	2	2	3
49	male	66	4	self	AMN	no	4.0	3.5	c.1386del (p.Ile463Serfs*95)	4	0	0	0	0	0	1	0	0	1	2	0	15	2	9	4	1	2	3	7
50	male	35	9	self	AMN	no	10.0	7.0	c.1825G>A (p.Glu609Lys)	3	0	0	0	0	0	1	0	0	1	1	0	7	6	0	1	0	0	1	0
51	female	37	5	mother	myeloneuropathy	no	1.5	n/a	c.311G>A (p.Arg104His)	2	1	0	0	0	0	0	0	0	1	0	0	22	1	0	21	4	2	4	11

Notes. Case numbers with the same superscript letter belong to the same family. ACALD = adult cerebral adrenoleukodystrophy; AMN = adrenomyeloneuropathy; AACS = adult ALD clinical score (range: 0.0-24.0, higher scores indicate greater neurological impairment); CCALD = childhood cerebral adrenoleukodystrophy; EDSS = Expanded Disability Status Scale (range: 0.0-10.0, higher scores indicate greater neurological impairment); PAFM = potentially affected family member.