Supplementary Material

Repeat-associated ataxias in a German patient cohort analysed by targeted parallel long-read sequencing

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Supplementary Table 1. Repeat-associated hereditary ataxias with their corresponding gene, location within the genome, tandem repeat sequence and thresholds for pathogenicity.

Disease	Gene	Genomic region	Repeat motif	Repeat sizes [RU] and their classification
Autosomal-dominant inheritance				
Spinocerebellar Ataxia 1 (SCA1)[1]	*ATXN1*	Exon	CAG	Normal: 6-35 Mutable normal: 36-38 [a] Pathogenic (full penetrance): > 38
SCA2[2]	*ATXN2*	Exon	CAG	Normal: ≤ 32 Pathogenic (reduced penetrance): 33-34 Pathogenic (full penetrance): > 34
SCA3[3]	*ATXN3*	Exon	CAG	Normal: < 44 Pathogenic (reduced penetrance): 45-51 Pathogenic (full penetrance): > 51
SCA4[b] [4,5]	*ZFHX3*	Exon	*GGC*	Normal: <49 Pathogenic (full penetrance): 48-57
SCA6[6]	*CACNA1A*	Exon	CAG	Normal: < 19 Pathogenic (reduced penetrance): 19 Pathogenic (full penetrance): 20-33
SCA7[7]	*ATXN7*	Exon	CAG	Normal: 7 – 27 Mutable normal: 28–33 Pathogenic (reduced penetrance): 34–36 Pathogenic (full penetrance): >36
SCA8[8]	*ATXN8OS/ATXN8*	3'-UTR	(CTA · TAG) (CTG · CAG)	Normal: 15–50 Pathogenic (incomplete penetrance): >50
SCA10[9]	*ATXN10*	Intron	ATTCT	Normal: 10-32 Reduced penetrance: 33-850 Full penetrance: >800
SCA12[10]	*PPP2R2B*	5'UTR	CAG	Normal: 7-28 intermediate: 33-50 Pathogenic (full penetrance): >50
SCA17[11]	*TBP*	Exon	CAG	Normal: 25-40 Pathogenic (reduced penetrance): 41-48 Pathogenic (full penetrance): 49-66
SCA27B[12–14]	*FGF14*	Intron	GAA	Normal: <180 intermediate alleles (unknown clinical significance): 180-249[15,16] Pathogenic (reduced penetrance): 250-300[c] Pathogenic (full penetrance): >300[c]
SCA31[17]	*BEAN1*	Intron	pathogenic: [(TGGAA)n(TAAAA)n(TAAAATAGAA)n] non-pathogenic: (TAAAA)$_{8-20}$ ((TAGAA)n(TAAAATAGAA)n] (TACAA)n	
SCA36[18]	*NOP56*	Intron	GGCCTG	Non-pathogenic: 3-14 Pathogenic: >650
SCA37[19]	*DAB1*	5'UTR	Non-pathogenic: (ATTTT)n Pathogenic: [(ATTTT)n(ATTTC)m(ATTTT)n], n>31; m = 31-75	

DRPLA[20]	*ATN1*	Exon	CAG	Normal: 6-35 Pathogenic (reduced penetrance): 35-47 Pathogenic (full penetrance): 48-93
Autosomal-recessive inheritance				
Friedreich ataxia (FRDA)[21]	*FXN*	Intron	GAA	Normal: 5-33 Pathogenic (reduced penetrance): 34-65 Pathogenic (full penetrance): 66-1700
RFC1 spectrum disorder (CANVAS)[d], [22–30]	*RFC1*	Intron	***(Likely) non-pathogenic:*** (AAAAG)$_{11}$ (AAAAG)$_{exp}$ (AAAGG)$_{<500\ \mathrm{RU}}$ (AAGGG)$_{<250\ \mathrm{RU}}$ (AAAGGG)$_{exp}$[e] (AAGAG)$_{exp}$ ***(Likely) pathogenic:*** (AAGGG)$_{>250\ \mathrm{RU}}$ (ACAGG)$_{exp}$ (AAAGG)$_{>500\ \mathrm{RU}}$ (AGGGC)$_{exp}$ (AGAGG)$_{exp}$[f] (AAGGC)$_{exp}$[f] ***Uncertain significance:*** (AGGGG)$_{exp}$ (ACAAG)$_{exp}$ (ACGGG)$_{complex}$[g]	
X-linked inheritance				
Fragile-X-associated tremor/ataxia syndrome (FXTAS)[31]	*FMR1*	5'-UTR	CGG	Normal: 5-54 Premutation allele (FXTAS-associated): 55–200 Full mutation (FXS-associated): >200

a If interrupted by CAT: normal alleles.

b Not analyzed in the primary cohort of 513 patients with adult-onset ataxia in this study.

c Only pure (GAA)$_n$ repeat expansions are considered as pathogenic.

d Clinical significance for several configurations is still controversially discussed. Classification is based on all available literature and might differ in some cases from individual reports. Configurations that were only detected in short-read genome data are not included.

e This repeat configuration exhibits a high degree of heterogeneity and is usually observed as a complex expansion carrying different motifs such as 'AAAGGG', 'AAGGG' and 'AAAGG'. In this and previous studies we therefore refer to this configuration as '(AAAGGG)$_{complex}$'.

f Only observed in isolated families.

g Similarly, to (AAAGGG)$_{complex}$ this repeat configuration showed a high level of heterogeneity with several motifs ('ACGGG', 'AAGGG'

Supplementary Table 2. CRISPR RNAs (crRNAs) to enrich repeat regions within the indicated loci.

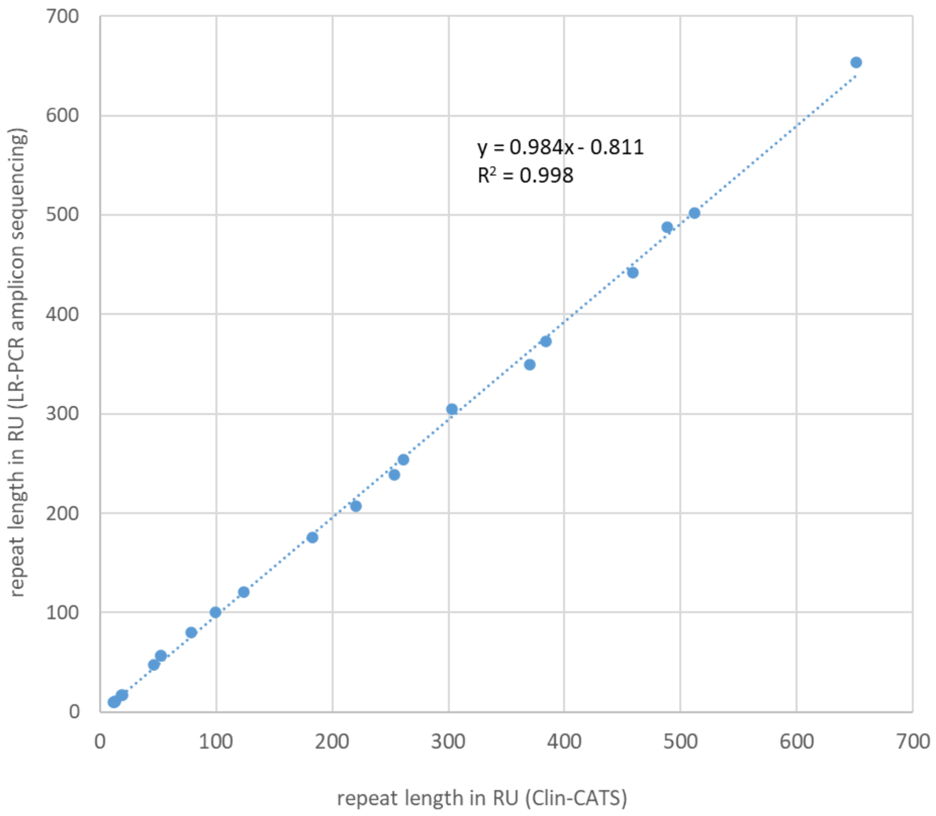
Locus	**Guide**	**Sequence 5'→3'**
ATN1	ATN1_D1	CGCTAGACGTTCTCGCTCCA
	ATN1_U2	GGGTGAGAAGTCTTAGTCGG
ATXN1	ATXN1_D1	GGTGGAAACTTTTATCGGTT
	ATXN1_U2	ATGTAATCGATCTAAGAACC
ATXN2	ATXN2_D1	GTCGGCTCTGTCTCTACCGA
	ATXN2_U2	CCGGTCACCCGCCGTCAAGC
ATXN3	ATXN3_D1	AGCGCATTCCCAAATAGACG
	ATXN3_U1	GATTACTGCTGAACGCACAT
CACNA1A	CACNA1A_D2	GGTCCAGTTCTGCGTGGAAT
	CACNA1A_U2	TTGGCACTCGGGCATAGACT
ATXN7	ATXN7_D1	ATCTAGGTTAAACTTCCCGC
	ATXN7_U2	CGGTACTTCGTCCTGACACC
ATXN8OS/ATXN8	ATXN8_D1	CATTACAGGTCACGCAAAGA
	ATXN8_U1	ATACTTGGCCATCGTAATTG
ATXN10	ATXN10_D2	CAGCACGTTGTTTACTGGAG
	ATXN10_U2	GTAGTTTCATCTCACTAAGA
PPP2R2B	PPP2R2B_D1	GAGATCTCATGAAGTACCGC
	PPP2R2B_U1	CCTCAGCTAAGTGTCGAAAT
	PPP2R2B_U2	TAACATCGTTTCGCTCAGGG
TBP	TBP_D1	GAGGTTACTACTGCATGTTG
	TBP_U1	TGAGACGAGTTCCAGCGCAA
BEAN1	BEAN1_D1	TGACCACCTCACCCGATGAA
	BEAN1_D2	AAGCATAGATCGCACCCAGTAGG
	BEAN1_U2	AAGTGACGCCCGCCCTCAGG
NOP56	NOP56_D2	ACTATCACAAGCTGCCCGAGAGG
	NOP56_D3	CTTCTGCTTTAATCGTTCGG
	NOP56_U1	AAATTTTTCCAGAGTACGACTGG
	NOP56_U4	TGTACTATTTGTCACAATCA
DAB1	DAB1_D2	GTTGGATGTGGGATACTCGA
	DAB1_U1	CTGGTTGAAACATGTCTCAG
FGF14	FGF14_D1	CTGCACTTGTCTGTTAATGA
	FGF14_U1	TAGATACTGTCATAGTAGGA
FXN	FXN_D1	CACCAGTTTCGAGAATCCTG
	FXN_U2	CTGCTGTAAACCCATACCGG
FMR1	FMR1_D2	ATCACGATCCCAATCTTCTC
	FMR1_U2	TTTAGGCTTGAGCAACGAAC
RFC1	RFC1_D1	TTCGTGGAACTATCTTGGTA
	RFC1_D2	TGATTACAACCATCAAGGAT
	RFC1_U1	TAACTTCCAACAACCTCAAC
	RFC1_U2	GCTCAGTCGTTTTAACCAGG
ZFHX3	ZFHX3_U1	TTTGAAGCTCCTACCAAACAAGG
	ZFXH3_D1	AACACCCTCTTGCATTACCTTGG

Supplementary Table 4. Comparison of median repeat lengths and repeat configurations determined by Clin-CATS and LR-PCR amplicon sequencing.

Pat.	**Clin-CATS**				**LR-PCR amplicon sequencing**			
	allele 1		**allele 2**		**allele 1**		**allele 2**	
	length[a] [RU]	**configuration**	**Length[a] [RU]**	**configuration**	**Length[a] [RU]**	**main motif**	**Length[a] [RU]**	**main motif**
195	18	(GAA)n	220	(GAA)n	17	GAA	207	GAA
215	99	(GAA)n	261	(GAA)n	100	GAA	254	GAA
246	52	$(GAA)_9(GAGGAA)_{12}(GAA)_{19}$	651	$(GAA)_6(GAAGGA)_{275}(GAA)_{95}$	57	GAA	653	GAAGGA
248	46	(GAA)n	459	(GAA)n	48	GAA	442	GAA
264	11	(GAA)n	370	(GAA)n	10	GAA	349	GAA
278	19	(GAA)n	183	(GAA)n	17	GAA	176	GAA
291	60	$(GAA)_8(GGA)_{28}(GAA)_{14}$	253	(GAA)n	-[b]	-[b]	239	GAA
299	12	(GAA)n	488	(GAA)n	10	GAA	488	GAA
311	78	(GAA)n	512	(GAA)n	80	GAA	502	GAA
350	124	(GAA)n	384	(GAA)n	121	GAA	373	GAA
364	13	(GAA)n	303	$(GAA)(GAAGGA)_{144}(GAA)_{14}$	11	GAA	305	GAAGGA

[a] Repeat length was calculated based on the assumptions of triplets (repeat size in bp divided by 3).

[b] LR-PCR gave only one product likely due to allelic drop-out.



Supplementary Figure 1. Scatterplot shows concordance of median repeat lengths determined by Clin-CATS and LR-PCR amplicon sequencing for the repeat in *FGF14*.

Supplementary Table 4. Genes and their OMIM analyzed within the ataxia next-generation-sequencing panel

Gene	OMIM
AAAS	605378
ABCB7	300135
ABHD12	613599
ADCY5	600293
AFG3L2	604581
AMACR	604489
AMPD2	102771
ANO10	613726
AP1S2	300629
APOB	107730
APTX	606350
ARG1	608313
ARMC9	617612
ARSA	607574
ASL	608310
ASS1	603470
ATCAY	608179
ATL1	606439
ATM	607585
ATP13A2	610513
ATP1A2	182340
ATP1A3	182350
ATP7B	606882
ATP8A2	605870
AUH	600529
B3GALNT2	610194
BCKDHA	608348
BCKDHB	248611
BICD2	609797
BRF1	604902
BSCL2	606158
C19orf12	614297
CA8	114815
CACNA1A	601011
CACNA1C	114205
CACNA1G	604065
CACNA2D2	607082
CACNB4	601949
CAMTA1	611501
CAPN1	114220
CASK	300172
CHCHD10	615903
CHMP1A	164010
CLCN2	600570
CLN6	606725
CLP1	608757
COA7	615623
COASY	609855
COG5	606821
COQ2	609825
COQ4	612898
COQ6	614647
COQ7	601683
COQ8A	606980
COQ9	612837
COX20	614698
CP	117700
CPS1	608307
CSTB	601145
CWF19L1	616120
CYP27A1	606530
CYP2U1	610670
DARS2	610956
DBT	248610
DCAF17	612515
DDHD2	615003
DNAJC19	608977
DNAJC5	611203
DNMT1	126375
EEF2	130610
EIF2B1	606686
EIF2B2	606454
EIF2B3	606273
EIF2B4	606687
EIF2B5	603945
ELOVL4	605512
EPM2A	607566
ERCC4	133520
ETFA	608053
ETFB	130410
ETFDH	231675
EXOSC3	606489
EXOSC8	606019
EXOSC9	606180
FA2H	611026
FGF14	601515
FLVCR1	609144
FMR1	309550
FOLR1	136430
FTL	134790
FXN	606829
GAN	605379
GBA2	609471
GBE1	607839
GFAP	137780
GJC2	608803
GLRA1	138491
GLRB	138492
GOSR2	604027
GPAA1	603048
GRID2	602368
GRM1	604473
HEXA	606869
HEXB	606873
HGSNAT	610453
HSD17B4	601860
HSPD1	118190
IBA57	615316
IRF2BPL	611720
ITPR1	147265
IVD	607036
KCNA1	176260
KCNA2	176262
KCNA4	176266
KCNC3	176264
KCND3	605411
KCNJ10	602208
KCNQ2	602235
KIF1A	601255
KIF1C	603060
KIF5A	602821
L1CAM	308840
LYST	606897
MARS2	609728
MFN2	608507
MID1	300552
MMACHC	609831
MRE11	600814
MSTO1	617619
MT-ATP6	516060
MTPAP	613669
MTRFR	613541
MTTP	157147
NHLRC1	608072
NIPA1	608145
NKX2-1	600635
NKX6-2	605955
NOP56	614154
NPC1	607623
NPC2	601015
NRAS	164790
OPA1	605290
OPA3	606580
OPHN1	300127
OTC	300461
PACS2	610423
PANK2	606157
PAX2	167409
PAX6	607108
PDHA1	300502
PDSS1	607429
PDSS2	610564
PDYN	131340
PEX16	603360
PEX6	601498
PEX7	601757
PHYH	602026
PLA2G6	603604
PLCB1	607120
PLP1	300401
PMPCA	613036
PMPCB	603131
PNKD	609023
PNKP	605610
PNPLA6	603197
POLG	174763
POLR3A	614258
PPP2R2B	604325
PRKCG	176980
PRNP	176640
PRRT2	614386
PTRH2	608625
PUM1	607204
RARS2	611524
REEP1	609139
RETREG1	613114
RFC1	102579
RNF170	614649
RNF216	609948
ROBO3	608630
RTN2	603183
RUBCN	613516
SACS	604490
SAMD9L	611170
SCN1A	182389
SCN2A	182390
SCN8A	600702
SEPSECS	613009
SETX	608465
SIL1	608005
SLC16A2	300095
SLC1A3	600111
SLC25A15	603861
SLC25A46	610826
SLC2A1	138140
SLC2A10	606145
SLC33A1	603690
SLC39A8	608732
SLC52A2	607882
SLC52A3	613350
SLC5A7	608761
SLC6A19	608893
SLC9A6	300231
SNX14	616105
SPAST	604277
SPG11	610844
SPG7	602783
SPR	182125
SPTBN2	604985
SQSTM1	601530
SRD5A3	611715
STUB1	607207
SYNE1	608441
SYNGAP1	603384
TACO1	612958
TERT	187270
TGM6	613900
TMEM240	616101
TOE1	613931
TPP1	607998
TSEN2	608753
TSEN54	608755
TTBK2	611695
TTC19	613814
TTPA	600415
TTR	176300
TUBA1A	602529
TUBB2B	612850
TUBB3	602661
TUBB4A	602662
TWNK	606075
UBA5	610552
UBQLN2	300264
UCHL1	191342
UPF3B	300298
VAMP1	185880
VLDLR	192977
VPS13D	608877
VPS53	615850
VRK1	602168
WASHC5	610657
WDR45	300526
WDR73	616144
WDR81	614218
WFS1	606201
WNK1	605232
WWOX	605131
XRCC1	194360
ZFYVE26	612012
ZFYVE27	610243

Supplementary Table 5. Family history, current age and diagnostic results of all patients with adult-onset ataxia analyzed by Clin-CATS (cohort 1).

Pat.	family history	Primary diagnosis	Secondary diagnosis	Additional findings (heterozygous carrier status, repeat expansions of unknown significance, alternative configurations in *FGF14*)	NGS sequencing analysis
1	negative	negative			negative
2	n.a.	negative		*FGF14*: 183 RU (GAA)n	not determined
3	positive	SCA27B (*FGF14*: 308 RU (GAA)n)			negative
4	negative	negative			SCA11 possible
5	positive	SCA2 (*ATXN2*: 36 RU)		*FGF14*: 228 RU (GAA)n	not determined
6	n.a.	*RFC1* spectrum disorder (*RFC1*: 720 RU (AAGGG)n /880 RU (AAGGG)n			not determined
7	positive	negative		carrier for *RFC1* spectrum disorder (*RFC1*: ((AAAAG)11/AAAGG)610)	SCA13
8	positive	negative			negative
9	n.a.	negative			not determined
10	n.a.	*RFC1* spectrum disorder (*RFC1*: (AAGGG)775/(AAGGG)785			not determined
11	n.a.	SCA27B (*FGF14*: 332 RU (GAA)n)			not determined
12	positive	*RFC1* spectrum disorder (*RFC1*: >720 RU (AAGGG)n/ >720 (AAGGG)n			not determined
13	negative	SCA27B (*FGF14*: 282 RU (GAA)n)			not determined
14	positive	negative			not determined
15	negative	negative			negative
16	negative	SCA27B (*FGF14*: 267 RU (GAA)n)			not determined
17	negative	*RFC1* spectrum disorder (*RFC1*: 700 RU (AAGGG)n/910 (AAGGG)n)		*FXN*: one allele of uncertain significance (GAA)6(GAAGGA)100	not determined
18	negative	SCA27B (*FGF14*: 294 RU (GAA)n)	SCA8 (*ATXN8*: 88 RU)	*FGF14*: 232 RU (GAA)n	not determined
19	positive	*RFC1* spectrum disorder (*RFC1*: >870 RU (AAGGG)n/>870 RU (AAGGG)n)			not determined
20	negative	negative			negative
21	positive	SCA3 (*ATXN3*: 70 RU)			negative
22	negative	negative			not determined
23	n.a.	SCA27B (*FGF14*: 302 RU (GAA)n)			not determined
24	negative	negative			not determined

25	positive	negative		not determined
26	negative	SCA27B (*FGF14*: 364 RU (GAA)n)		negative
27	negative	negative		not determined
28	positive	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)105/(AAGGG)528)	negative
29	n.a.	SCA27B (*FGF14*: 356 RU (GAA)n)	*FGF14*: 189 RU (GAA)n	not determined
30	positive	negative	*FGF14*: 193 RU (GAA)n	negative
31	n.a.	*RFC1* spectrum disorder (*RFC1*: >890 RU (AAGGG)n/>890 RU (AAGGG)n)	carrier for FRDA (*FXN*: 91 RU (GAA)n))	negative
32	n.a.	DRPLA (*ATN1*: 50 RU)	*FGF14*: 180 RU (GAA)n	not determined
33	negative	negative		not determined
34	n.a.	*RFC1* spectrum disorder (*RFC1*: 700 RU (AAGGG)n/1290 RU (AAGGG)n)	*FGF14*: 200 RU (GAA)n	not determined
35	positive	SCA27B (*FGF14*: 389 RU (GAA)n)		negative
36	negative	SCA8 (*ATXN8*: 80 RU)		not determined
37	negative	negative	*FGF14*: 219 RU (GAA)n	not determined
38	negative	negative		negative
39	negative	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)1030)	negative
40	negative	negative		negative
41	n.a.	SCA27B (*FGF14*: 289 RU (GAA)n)		not determined
42	n.a.	SCA8 (*ATXN8*; 105 RU/140 RU (biallelic expansion))		not determined
43	negative	SCA27B (*FGF14*: 366 RU (GAA)n)		not determined
44	n.a.	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)730)	negative
45	positive	negative		negative
46	negative	SCA27B (*FGF14*: 271 RU (GAA)n)		not determined
47	negative	negative	*RFC1*: one allele of uncertain significance (AAAGG)317(AAGGG)210 (other allele: wildtype)	SPG39 possible
48	positive	SCA8 (*ATXN8*: 55 RU)	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)873)	negative
49	n.a.	negative		not determined
50	negative	SCA27B (*FGF14*: 266 RU (GAA)n)		negative
51	positive	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)904)	negative

52	positive	negative		negative
53	negative	negative		not determined
54	n.a.	negative	*FGF14*:(GAAGGA)4(GGA)168	not determined
55	negative	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)696)	SCA11
56	positive	SCA27B (*FGF14*: 267 RU (GAA)n)		negative
57	negative	negative		negative
58	negative	negative		not determined
59	positive	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)520)	negative
60	negative	negative		not determined
61	positive	negative		negative
62	positive	negative		EA2
63	positive	negative		negative
64	negative	*RFC1* spectrum disorder (*RFC1*: >1000 RU (AAGGG)n/>1000 RU (AAGGG)n)		not determined
65	positive	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)409)	negative
66	n.a.	negative		not determined
67	n.a.	SCA3 (*ATXN3*: 66 RU)		negative
68	negative	negative		negative
69	n.a.	negative		not determined
70	negative	*RFC1* spectrum disorder (*RFC1*: >800 RU (AAGGG)n/>800 RU (AAGGG)n)		not determined
71	positive	negative		not determined
72	negative	negative		negative
73	negative	SCA27B (*FGF14*: 425 RU (GAA)n)		not determined
74	positive	*RFC1* spectrum disorder (*RFC1*: >990 RU (AAGGG)n/>990 RU (AAGGG)n)		not determined
75	n.a.	FXTAS (*FMR1*: 56 RU, male)		not determined
76	positive	SCA36 (*NOP56*: >650 RU)		negative
77	negative	negative		negative
78	negative	negative		not determined
79	positive	SCA27B (*FGF14*: 330 RU (GAA)n)		not determined

80	positive	negative		not determined
81	negative	SCA27B (*FGF14*: 295 RU (GAA)n)		negative
82	negative	negative		negative
83	n.a.	*RFC1* spectrum disorder (*RFC1*: >600 RU (AAGGG)n/>600 RU (AAGGG)n)		not determined
84	negative	negative		POLR3A-associated ataxia
85	n.a.	negative		SCAR8
86	negative	SCA8 (*ATXN8*: 1230 RU)		not determined
87	positive	SCA3 (*ATXN3*: 72 RU)		not determined
88	positive	*RFC1* spectrum disorder (*RFC1*: 905 RU (AAGGG)n/905 RU (AAGGG)n)		not determined
89	negative	negative		negative
90	n.a.	negative		negative
91	negative	negative		not determined
92	negative	negative		not determined
93	positive	negative		negative
94	positive	negative		not determined
95	n.a.	negative		not determined
96	n.a.	*RFC1* spectrum disorder (*RFC1*: 620 RU (AAGGG)n/950 RU (AAGGG)n)		not determined
97	negative	negative		not determined
98	n.a.	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)115/(AAGGG)931)	negative
99	negative	SCA27B (*FGF14*: 288 RU (GAA)n)		not determined
100	negative	SCA27B (*FGF14*: 352 RU (GAA)n)		negative
101	negative	negative		negative
102	negative	*RFC1* spectrum disorder (*RFC1*: 554 RU (AAGGG)n/1395 RU (AAGGG)n)		not determined
103	negative	negative	*FGF14*: 239 RU (GAA)n	negative
104	negative	negative	*RFC1*: (AAAAG)11/(AAGGG)202	negative
105	n.a.	SCA27B (*FGF14*: 335 RU (GAA)n)		negative
106	negative	*RFC1* spectrum disorder (*RFC1*: 265 RU (AAGGG)n/1280 RU (AAGGG)n)	*FGF14*: 233 RU (GAA)n	not determined
107	n.a.	*RFC1* spectrum disorder (*RFC1*: 740 RU (AAGGG)n/830 RU (AAGGG)n)		not determined

108	positive	SCA27B (*FGF14*: 345 RU (GAA)n)			not determined
109	n.a.	negative		carrier for FRDA (*FXN*: 93 RU)	negative
110	negative	negative			SCA42 possible
111	negative	negative		*FGF14*: 385 RU (GAAGGA)n	negative
112	negative	negative			not determined
113	n.a.	negative			not determined
114	negative	negative			negative
115	n.a.	SCA27B (*FGF14*: 313 RU (GAA)n)			not determined
116	n.a.	SCA27B (*FGF14*: 630 RU (GAA)n)			negative
117	negative	negative			not determined
118	positive	*RFC1* spectrum disorder (*RFC1*: 955 RU (AAGGG)n/1340 RU (AAGGG)n)			not determined
119	negative	negative			negative
120	n.a.	negative			not determined
121	negative	SCA27B (*FGF14*: 257 RU (GAA)n)		carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)100/(AAGGG)344)	negative
122	negative	SCA27B (*FGF14*: 279 RU (GAA)n)			negative
123	negative	negative			not determined
124	negative	negative			not determined
125	n.a.	negative			negative
126	negative	SCA27B (*FGF14*: 265 RU (GAA)n)	SCA17 (*TBP*: 44 RU)		negative
127	negative	negative			SPG79A
128	positive	*RFC1* spectrum disorder (*RFC1*: 784 RU (AAGGG)n/1300 RU (AAGGG)n)			not determined
129	n.a.	negative			not determined
130	positive	negative		carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)1128)	negative
131	n.a.	*RFC1* spectrum disorder (*RFC1*: 848 RU (AAGGG)n/1090 RU (AAGGG)n)			not determined
132	negative	negative		carrier for FRDA (*FXN*: 827 RU)	negative
133	negative	SCA1 (*ATXN1*: 50 RU)			not determined
134	n.a.	negative			not determined
135	n.a.	negative			not determined
136	negative	negative			not determined

137	positive	SCA6 (*CACNA1A*: 23 RU)		not determined
138	positive	negative		ataxia telangiectasia
139	n.a.	negative		negative
140	n.a.	SCA27B (*FGF14*: 372 RU (GAA)n)		negative
141	positive	*RFC1* spectrum disorder (*RFC1*: (AAGGG)710/(AAGGG)>900)		not determined
142	n.a.	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)830)	negative
143	n.a.	negative		Developmental and epileptic encephalopathy
144	positive	SCA27B (*FGF14*: 286 RU (GAA)n)	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)500)	negative
145	positive	negative	*FGF14*: 181 RU (GAA)n	not determined
146	n.a.	negative		negative
147	negative	negative		negative
148	n.a.	*RFC1* spectrum disorder (*RFC1*: (AAGGG)799/(AAGGG)1200)		not determined
149	positive	negative		negative
150	negative	negative	*FGF14*: 193 RU (GAA)n	not determined
151	n.a.	negative		EA2
152	n.a.	SCA6 (*CACNA1A*: 22 RU)		not determined
153	negative	SCA1 (*ATXN1*: 49 RU)		not determined
154	positive	SCA27B (*FGF14*: 610 RU (GAA)n)		negative
155	positive	*RFC1* spectrum disorder (*RFC1*: (AAGGG)753/(AAGGG)963)		not determined
156	n.a.	negative	*FGF14*: 219 RU (GAA)n	not determined
157	negative	SCA3 (*ATXN3*: 64 RU)		not determined
158	n.a.	negative		negative
159	negative	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAAGG)590)	negative
160	negative	negative		not determined
161	negative	negative		not determined
162	n.a.	negative		not determined
163	negative	negative		negative
164	negative	negative		not determined

165	negative	negative		negative
166	positive	negative		SCA28 possible
167	n.a.	SCA3 (*ATXN3*: 78 RU)	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)589)	negative
168	negative	negative		not determined
169	positive	negative		negative
170	n.a.	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)131/(AAAGG)610)	negative
171	n.a.	negative		not determined
172	n.a.	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)58(AAGAG)32/(AAGGG)840)	negative
173	negative	SCA8 (*ATXN8*: 110 RU)	*FGF14*: 230 RU (GAA)n	not determined
174	n.a.	negative	*FGF14*: 174 RU (GAAGGA)n	not determined
175	negative	SCA8 (*ATXN8*: 250 RU)		not determined
176	negative	SCA27B (*FGF14*: 325 RU (GAA)n)	*RFC1*: (AAAAG)11/(AAGGG)83	not determined
177	n.a.	SCA27B (*FGF14*: 472 RU (GAA)n)		not determined
178	negative	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)755)	negative
179	negative	negative		negative
180	negative	negative		not determined
181	n.a.	SCA37 (*DAB1*: (ATTTT)155(ATTTC)100(ATTTT)10)		not determined
182	n.a.	negative	*FGF14*: 235 RU (GAA)n	not determined
183	negative	negative		not determined
184	negative	negative		negative
185	n.a.	negative		not determined
186	n.a.	negative		not determined
187	n.a.	negative		negative
188	n.a.	negative		not determined
189	negative	negative		negative
190	n.a.	*RFC1* spectrum disorder (*RFC1*: (AAGGG)>850/(AAGGG)>850		not determined
191	negative	negative		negative
192	n.a.	negative		negative

193	negative	negative			not determined
194	negative	negative			negative
195	negative	negative		*FGF14*: 220 RU (GAA)n	not determined
196	negative	negative		*FGF14*: 181 RU (GAA)n	not determined
197	negative	SCA27B (*FGF14*: 328 RU (GAA)n)			not determined
198	positive	negative			not determined
199	negative	negative			not determined
200	n.a.	negative			not determined
201	negative	negative		carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)620)	negative
202	negative	negative		*FGF14*: 199 RU (GAA)n; carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)689)	SPG7
203	n.a.	negative			negative
204	negative	negative		*RFC1*: (AAAGG)94/(AAGGG)220	not determined
205	n.a.	negative		*FGF14*: 222 RU (GAA)n	not determined
206	negative	negative	carrier for FRDA (*FXN*: 956 RU (GAA)n/normal-size allele)	*FGF14*: 181 RU (GAA)n	negative
207	negative	FXTAS (*FMR1*: 56 RU, female)			negative
208	negative	FXTAS (*FMR1*: 93 RU, male)			not determined
209	negative	negative			not determined
210	n.a.	negative		*FGF14*: 194 RU (GAA)n	not determined
211	negative	negative		*FGF14*: 212 RU (GAA)n	not determined
212	n.a.	negative			not determined
213	n.a.	negative			negative
214	n.a.	negative			negative
215	positive	SCA27B (*FGF14*: 261 RU (GAA)n)			not determined
216	n.a.	*RFC1* spectrum disorder (*RFC1*: (AAGGG)1065/(AAAGG)745			not determined
217	n.a.	negative			not determined
218	negative	negative			not determined
219	n.a.	negative			not determined
220	negative	negative			negative

221	negative	SCA27B (*FGF14*: 292 RU (GAA)n)		not determined
222	n.a.	negative		negative
223	n.a.	negative		not determined
224	positive	negative		not determined
225	n.a.	negative		not determined
226	n.a.	SCA27B (*FGF14*: 316 RU (GAA)n)		not determined
227	negative	negative		not determined
228	n.a.	negative		not determined
229	n.a.	negative		not determined
230	n.a.	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)660)	negative
231	positive	negative	*FGF14*: (GGA)75(GAA)300	not determined
232	n.a.	SCA27B (*FGF14*: 325 RU (GAA)n)		not determined
233	positive	negative		SCAR10
234	negative	negative		negative
235	negative	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)343)	negative
236	n.a.	negative		not determined
237	negative	negative		not determined
238	negative	negative		negative
239	positive	negative		not determined
240	n.a.	negative		not determined
241	n.a.	SCA8 (*ATXN8*: 100 RU)		not determined
242	positive	SCA1 (*ATXN1*: 51 RU)		not determined
243	positive	negative	*FGF14*: 173 RU (GAAGGA)n	negative
244	n.a.	negative		not determined
245	negative	negative	*FGF14*: 142 RU (GAAGGA)n	not determined
246	negative	negative	*FGF14*: (GAA)6(GAAGGA)275(GAA)95	not determined
247	n.a.	negative		not determined
248	negative	SCA27B (*FGF14*: 459 RU (GAA)n)		not determined
249	positive	negative		not determined

250	positive	negative		negative
251	negative	negative		not determined
252	negative	*RFC1* spectrum disorder (*RFC1*: (AAGGG)705/(AAGGG)>800)		negative
253	n.a.	negative		not determined
254	negative	negative		not determined
255	n.a.	negative		not determined
256	negative	negative		negative
257	n.a.	negative		NARP syndrome
258	n.a.	SCA17 (*TBP*: 43 RU)	*FGF14*: (GAA)2(GAAGGA)133(GAA)19	SCA48 possible
259	negative	negative		negative
260		negative		negative
261	negative	negative		SCA14 possible
262	n.a.	negative		negative
263	negative	FRDA (*FXN*: 100 RU/900 RU)		not determined
264	negative	SCA27B (*FGF14*: 370 RU (GAA)n)		negative
265	n.a.	negative		not determined
266	n.a.	negative		not determined
267	n.a.	negative		not determined
268	positive	negative		not determined
269	positive	*RFC1* spectrum disorder (*RFC1*: (AAGGG)868/(AAGGG)1132)		not determined
270	n.a.	negative		not determined
271	negative	negative		not determined
272	negative	SCA8 (*ATXN8*: 233 RU)		not determined
273	negative	negative		negative
274	negative	*RFC1* spectrum disorder (*RFC1*: (AAGGG)868/(AAGGG)1132)		negative
275	positive	negative		not determined
276	n.a.	negative		not determined
277	n.a.	negative		not determined
278	n.a.	negative	*FGF14*: 183 RU (GAA)n	not determined

279	positive	SCA6 (*CACNA1A*: 22 RU)			not determined
280	n.a.	SCA27B (*FGF14*: 307 RU (GAA)n)			not determined
281	negative	negative			not determined
282	n.a.	negative			not determined
283	n.a.	negative			not determined
284	negative	FXTAS (*FMR1*: 113 RU, male)			not determined
285	n.a.	SCA27B (*FGF14*: 414 RU (GAA)n)			not determined
286	n.a.	negative			negative
287	n.a.	negative			not determined
288	positive	SCA27B (*FGF14*: 427 RU (GAA)n)			not determined
289	positive	negative		*FGF14*: 180 RU (GAA)n	negative
290	n.a.	negative			not determined
291	negative	SCA27B (*FGF14*: 253 RU (GAA)n)			negative
292	n.a.	negative			not determined
293	n.a.	negative			negative
294	n.a.	*RFC1* spectrum disorder (*RFC1*: (AAGGG)>650/(AAGGG)>650)			not determined
295	negative	negative			not determined
296	positive	negative			negative
297	n.a.	FRDA (*FXN*: 567 RU/852 RU)	SCA17 (*TBP*: 47 RU)		not determined
298	positive	SCA8 (*ATXN8*: 80 RU)		*FGF14*: 219 RU (GAA)n	not determined
299	n.a.	SCA27B (*FGF14*: 488 RU (GAA)n)			negative
300	n.a.	negative			not determined
301	n.a.	negative			not determined
302	n.a.	negative			not determined
303	n.a.	negative			not determined
304	n.a.	negative			not determined
305	positive	SCA27B (*FGF14*: 293 RU (GAA)n)			not determined
306	n.a.	negative			not determined
307	n.a.	negative			not determined
308	n.a.	negative			not determined

309	negative	negative		negative
310	positive	SCA10 (*ATXN10*: (ATTCT)140(ATTCC)1325)	*FXN*: (GAA)10(GAAGGA(47)GAA(8)	negative
311	negative	SCA27B (*FGF14*: 512 RU (GAA)n)		not determined
312	positive	negative		negative
313	positive	negative		SCA42
314	n.a.	negative		not determined
315	n.a.	negative		negative
316	negative	negative		negative
317	positive	SCA27B (*FGF14*: 253 RU (GAA)n)		SPG5A
318	negative	*RFC1* spectrum disorder (*RFC1*: (AAGGG)>610/(AAGGG)>610)		not determined
319	n.a.	negative		negative
320	positive	negative		negative
321	n.a.	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)748)	negative
322	positive	negative		negative
323	positive	SCA27B (*FGF14*: 423 RU (GAA)n)		not determined
324	n.a.	negative		not determined
325	n.a.	negative		not determined
326	positive	negative		not determined
327	n.a.	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAGG)360/(AAGGG)556)	negative
328	n.a.	SCA6 (*CACNA1A*: 23 RU)		not determined
329	n.a.	SCA27B (*FGF14*: 314 RU (GAA)n)	*FXN*: (GAA)13(GAAGAG)17(GAA)23	not determined
330	negative	FRDA (*FXN*: 781 RU/160 RU)		not determined
331	n.a.	negative		not determined
332	negative	negative		not determined
333	n.a.	negative		negative
334	n.a.	negative		not determined
335	n.a.	SCA2 (*ATXN2*: 32 RU)		not determined
336	n.a.	SCA27B (*FGF14*: 340 RU (GAA)n)		not determined
337	n.a.	SCA27B (*FGF14*: 340 RU (GAA)n)	*FGF14*: 220 RU (GAA)n	not determined

338	negative	negative		not determined
339	n.a.	negative		not determined
340	n.a.	negative		negative
341	n.a.	negative		not determined
342	n.a.	negative		negative
343	n.a.	SCA27B (*FGF14*: 262 RU (GAA)n)		negative
344	n.a.	negative		not determined
345	n.a.	negative		negative
346	negative	SCA3 (*ATXN3*: 74 RU)		not determined
347	negative	negative		negative
348	positive	SCA27B (*FGF14*: 370 RU (GAA)n)		not determined
349	positive	negative		not determined
350	negative	SCA27B (*FGF14*: 384 RU (GAA)n)		not determined
351	negative	negative		not determined
352	n.a.	negative		not determined
353	n.a.	negative		not determined
354	n.a.	SCA27B (*FGF14*: 285 RU (GAA)n)		not determined
355	n.a.	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAAGG)526)	negative
356	n.a.	negative		not determined
357	n.a.	negative		not determined
358	negative	negative		negative
359	n.a.	negative	*FXN*: (GAA)6(GAA interrupted by GAG)69(GAA)15	not determined
360	n.a.	*RFC1* spectrum disorder (*RFC1*: (AAGGG)>695/(AAGGG)>695)		not determined
361	negative	negative		not determined
362	n.a.	negative		negative
363	n.a.	negative		not determined
364	n.a.	negative	*FGF14*: (GAA)4(GAAGGA)144(GAA)14	not determined
365	negative	negative		not determined
366	negative	negative		not determined

367	positive	SCA27B (*FGF14*: 353 RU (GAA)n)		not determined
368	positive	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)325)	negative
369	n.a.	SCA27B (*FGF14*: 410 RU (GAA)n)		not determined
370	n.a.	negative		not determined
371	negative	negative		not determined
372	n.a.	negative		not determined
373	n.a.	negative	*FGF14*: 224 RU (GAA)n	not determined
374	negative	negative		not determined
375	negative	negative		SCA42 possible
376	n.a.	negative	*FGF14*: 237 RU (GAA)n	not determined
377	negative	*RFC1* spectrum disorder (*RFC1*: (AAGGG)680/(AAGGG)680)		not determined
378	n.a.	negative		not determined
379	negative	SCA27B (*FGF14*: 395 RU (GAA)n)		not determined
380	n.a.	negative		not determined
381	n.a.	negative	*FGF14*: 185 RU (GAAGGA)n	not determined
382	n.a.	negative		negative
383	negative	negative		not determined
384	negative	negative	*FGF14*: 217 RU (GAA)n	not determined
385	n.a.	negative		not determined
386	n.a.	negative		not determined
387	n.a.	negative		negative
388	n.a.	negative		not determined
389	positive	negative		not determined
390	n.a.	negative		not determined
391	positive	negative		not determined
392	n.a.	negative		not determined
393	negative	negative		Tay-Sachs disease likely
394	positive	negative		SPG7
395	positive	negative		not determined

396	n.a.	SCA27B (*FGF14*: 364 RU (GAA)n)		carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)695)	negative
397	positive	negative			not determined
398	positive	negative			not determined
399	n.a.	negative			not determined
400	positive	negative			not determined
401	n.a.	negative			not determined
402	n.a.	negative			not determined
403	positive	negative			not determined
404	negative	SCA8 (*ATXN8*: 92 RU)		*FGF14*: (GAA)3(GAAGGA)131(GAA)18	not determined
405	n.a.	SCA27B (*FGF14*: 310 RU (GAA)n)			not determined
406	negative	negative			not determined
407	positive	negative			not determined
408	n.a.	*RFC1* spectrum disorder (*RFC1*: (AAGGG)>890/(AAGGG)>890)			not determined
409	negative	SCA27B (*FGF14*: 331 RU (GAA)n)	FXTAS (*FMR1*: 58 RU)	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAAGG)517)	negative
410	positive	*RFC1* spectrum disorder (*RFC1*: (AAAGG)776/(AAGGG)750)			not determined
411	n.a.	SCA27B (*FGF14*: 310 RU (GAA)n)			not determined
412	n.a.	negative		*FGF14*: 276 RU (GAAGGA)n;	negative
413	n.a.	negative			not determined
414	n.a.	negative			not determined
415	n.a.	SCA27B (*FGF14*: 292 RU (GAA)n)			negative
416	positive	negative			not determined
417	n.a.	SCA27B (*FGF14*: 359 RU (GAA)n)			not determined
418	n.a.	negative			not determined
419	negative	SCA27B (*FGF14*: 292 RU (GAA)n)		*FGF14*: 238 RU (GAA)n	not determined
420	negative	negative			negative
421	negative	negative			not determined
422	n.a.	*RFC1* spectrum disorder (*RFC1*: (AAGGG)633/(AAGGG)1012)			not determined
423	negative	SCA1 (*ATXN1*: 43 RU)			not determined
424	n.a.	SCA8 (*ATXN8*: 90 RU)			not determined

425	n.a.	negative			not determined
426	n.a.	negative			not determined
427	positive	SCA27B (*FGF14*: 344 RU (GAA)n)		*RFC1*: (AAAAG)11/(AAGGG)227	not determined
428	n.a.	negative		*FGF14*: 169 RU (GAAGGA)n	negative
429	n.a.	negative			not determined
430	positive	SCA10 (*ATXN10*: (ATTCT)120(ATTCC)1450)		*FXN*: (GAA)10(GAAGGA)47(GAA)8	not determined
431	negative	negative			not determined
432	negative	SCA27B (*FGF14*: 252 RU (GAA)n	FXTAS (*FMR*): 57 RU)		negative
433	n.a.	*RFC1* spectrum disorder (*RFC1*: (AAGGG)>980/(AAGGG)>980)			not determined
434	positive	SCA27B (*FGF14*: 469 RU (GAA)n)			not determined
435	negative	negative			not determined
436	positive	negative			not determined
437	negative	negative		*FGF14*: 242 RU (GAA)n	negative
438	n.a.	negative			negative
439	n.a.	SCA27B (*FGF14*: 341 RU (GAA)n)		carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)755)	negative
440	negative	negative			not determined
441	positive	SCA3 (*ATXN3*: 75 RU)			not determined
442	n.a.	SCA27B (*FGF14*: 309 RU (GAA)n)			not determined
443	n.a.	negative		carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)398)	negative
444	n.a.	SCA27B (*FGF14*: 265 RU (GAA)n)		*FGF14*: 220 RU (GAA)n	not determined
445	n.a.	SCA27B (*FGF14*: 298 RU (GAA)n)			not determined
446	n.a.	negative			not determined
447	n.a.	SCA27B (*FGF14*: 352 RU (GAA)n)			not determined
448	n.a.	negative			not determined
449	negative	SCA27B (*FGF14*: 296 RU (GAA)n)			not determined
450	n.a.	*RFC1* spectrum disorder (*RFC1*: (AAGGG)>620/(AAGGG)>620)			not determined
451	n.a.	SCA27B (*FGF14*: 251 RU (GAA)n)			not determined
452	negative	negative			negative
453	negative	negative			negative

454	n.a.	negative		not determined
455	negative	FXTAS (*FMR1*: 95 RU, male)	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)343)	negative
456	negative	negative		not determined
457	n.a.	negative		not determined
458	n.a.	negative		not determined
459	n.a.	negative		not determined
460	negative	negative		negative
461	negative	SCA6 (*CACNA1A*: 22 RU)		not determined
462	positive	negative	*FGF14*: ((GAA)58(GAAGGA)192(GAA)56)	negative
463	negative	negative		not determined
464	n.a.	*RFC1* spectrum disorder (*RFC1*: (AAGGG)>400/(AAGGG)>400)		negative
465	n.a.	negative	*FGF14*: 225 RU (GAA)n	not determined
466	positive	SCA27B (*FGF14*: 352 RU (GAA)n)		not determined
467	n.a.	negative		negative
468	negative	negative		negative
469	n.a.	SCA27B (*FGF14*: 313 RU (GAA)n)		not determined
470	negative	negative		not determined
471	negative	negative		not determined
472	n.a.	negative		not determined
473	negative	negative		negative
474	n.a.	negative		not determined
475	n.a.	negative		negative
476	n.a.	negative		negative
477	n.a.	negative		negative
478	positive	negative	*FGF14*: 230 RU (GAA)n	negative
479	n.a.	negative		negative
480	negative	negative		negative
481	positive	SCA27B (*FGF14*: 326 RU (GAA)n)	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)730)	negative
482	positive	negative		not determined

483	n.a.	*RFC1* spectrum disorder (*RFC1*: (AAGGG)>620/(AAGGG)>620)		not determined
484	negative	negative		negative
485	n.a.	negative		negative
486	positive	*RFC1* spectrum disorder (*RFC1*: (AAGGG)>800/(AAGGG)>800)		not determined
487	n.a.	negative		not determined
488	negative	negative		not determined
489	n.a.	negative		not determined
490	negative	negative		not determined
491	n.a.	negative		not determined
492	n.a.	negative		not determined
493	n.a.	FXTAS (*FMR1*: 85 RU, male)		not determined
494	n.a.	negative	*FGF14*: 346 RU (GAAGGA)n	negative
495	positive	negative	*RFC1*: (AAAAG)11/(AAGGG)214	not determined
496	n.a.	negative		negative
497	positive	negative	carrier for *RFC1* spectrum disorder (*RFC1*: (AAAAG)11/(AAGGG)608)	negative
498	negative	negative		negative
499	negative	SCA27B (*FGF14*: 296 RU, (GAA)n)		not determined
500	n.a.	negative	*FGF14*: 183 RU (GAA)n	negative
501	n.a.	negative		not determined
502	n.a.	*RFC1* spectrum disorder (*RFC1*: (AAGGG)>630/(AAGGG)>630)		not determined
503	n.a.	*RFC1* spectrum disorder (*RFC1*: (AAGGG)655/(AAGGG)825)		not determined
504	n.a.	negative		not determined
505	negative	SCA27B (*FGF14*: 320 RU (GAA)n)		not determined
506	negative	FXTAS (*FMR1*: 93 RU, male)		not determined
507	n.a.	negative	carrier for FRDA (*FXN*: 915 RU)	negative
508	n.a.	*RFC1* spectrum disorder (*RFC1*: (AAGGG)>700/(AAGGG)>700)		not determined
509	n.a.	SCA27B (*FGF14*: 308 RU, (GAA)n)		not determined
510	postive	SCA27B (*FGF14*: 324 RU, (GAA)n)		not determined

511	n.a.	SCA27B (*FGF14*: 327 RU, (GAA)n)		not determined
512	negative	SCA27B (*FGF14*: 351 RU, (GAA)n)		not determined
513	n.a.	SCA6: (*ATXN6*: 20 RU homozygous)	*CACNA1A*: biallelic expansion	not determined

Supplementary Table 6. Age of onset and frequency of different symptoms in patients with primary diagnosis of SCA1–3, 6, 8, 27B, FXTAS, *RFC1* spectrum disorder. In the headline total number of patients with primariy diagnosis of the respective condition is given. For DRPLA, SCA17, 36, 37 and FRDA findings are only discussed in the manuscript as only a few patients each were identified.

SCA1 (n = 4)		
Age-of-onset	44.5 years (n=4); range 36–54 years	
Symptom	**Affected patients/patients with information available**	**Frequency [%]**
cerebellar oculomotor signs	3/3	100
gait ataxia	4/4	100
limb ataxia	3/3	100
dysarthria	3/3	100
neuropathy	2/3	67
other non-cerebellar signs[a]	1/2	50
reported symptoms (number of patients):	hyperreflexia (1)	
SCA2 (n = 2)		
Age-of-onset	47, 62 (n=2)	
Symptom	**Affected patients/patients with information available**	
cerebellar oculomotor signs	1/2	50
gait ataxia	2/2	100
limb ataxia	2/2	100
dysarthria	1/2	50
neuropathy	2/2	100
other non-cerebellar signs[a]	1/2	50
reported symptoms (number of patients):	spasticity (1)	
SCA3 (n = 7)		
Age-of-onset	49 years (n=5); range: 46–62 years	

Symptom	**Affected patients/patients with information available**	
cerebellar oculomotor signs	4/4	100
gait ataxia	5/5	100
limb ataxia	5/5	100
dysarthria	2/3	67
neuropathy	3/3	100
other non-cerebellar signs[a]	2/2	100
reported symptoms (number of patients):	spasticity (1); BVP and restless legs syndrome (1)	
SCA6 (n = 6)		
Age-of-onset	66 years (n=4), range: 65–70 years	
Symptom	Affected patients/patients with information available	Frequency [%]
cerebellar oculomotor signs	4/4	100
gait ataxia	5/5	100
limb ataxia	2/3	67
dysarthria	3/3	100
neuropathy	2/4	50
other non-cerebellar signs	1/3	
reported symptoms (number of patients):	pyramidal sign (1)	
SCA8 (n = 11)		
Age-of-onset	55 years (n=7), range 38–71 years	
Symptom	Affected patients/patients with information available	Frequency [%]
cerebellar oculomotor signs	7/8	88
gait ataxia	8/9	89
limb ataxia	6/7	88

dysarthria	5/7	71
neuropathy	1/8	13
other non-cerebellar signs	6/9	67
reported symptoms (number of patients):	Hyperkinesia (2), upper motor neuron signs (5), hypokinesia (1), autonomic dysfunction (1), REM sleep disorder (1)	
SCA27B (n = 83, > 250 RU)		
Age-of-onset	62 years (n=63); range 35–82 years	
Symptom	Affected patients/patients with information available	Frequency [%]
episodic onset	30/58	52
cerebellar oculomotor signs	55/60	92
downbeat nystagmus	27/50	54
gait ataxia	72/76	95
limb ataxia	35/53	66
dysarthria	32/60	53
neuropathy	24/56	43
sensory (clinical diagnosis)	8/26	
sensorimotor (clinical diagnosis)	2/26	
axonal sensory	2/26	
demyelinating sensory	1/26	
axonal sensorimotor	5/26	
not classified	6/26	
bilateral vestibulopathy	17/40	43
autonomic dysfunction	3/49	6

other non-cerebellar signs	14/50	28
reported symptoms (number of patients):	upper motor neuron signs (8), lower motor neuron signs (2), parkinsonism (2), hyperkinesia (1), chorea (1)	
Combination of symptoms		
neuropathy and cerebellar ataxia	7/40	
neuropathy, cerebellar ataxia, bilateral vestibulopathy (CANVAS triad)	10/40	
***FXTAS* (n = 7; six males, one female (no data available))**		
Age-of-onset	66 years (n=5); range 57–73 years	
Symptom	Affected patients/patients with information available	Frequency [%]
cerebellar oculomotor signs	2/4	50
gait ataxia	4/5	80
limb ataxia	4/5	80
dysarthtria	3/6	50
neuropathy	3/5	60
tremor	3/5	60
parkinsonism	2/5	40
cognitive impairment (executive dysfunction, memory deficiency)	2/5	40
white matter lesion in middle cerebellar peduncles (MCP sign)	3/5	60
propability of FXTAS according to clinical and radiological criteria		
definite diagnosis of FXTAS according to ref. 21	3/6	
probable diagnosis of FXTAS according to ref. 21	2/6	
possible diagnosis of FXTAS according to ref. 21	0/6	
not evaluated due to incomplete data	1/6 (female *FMR1* premutation carrier)	
***RFC1* spectrum disorder (n = 83, > 250 RU)**		

Age-of-onset	54.5 years (n=28); range 41–78 years	
Symptom	Affected patients/patients with information available	Frequency [%]
cerebellar oculomotor signs	27/30	90
gait ataxia	31/31	100
limb ataxia	25/28	89
dysarthria	14/25	56
neuropathy	32/32	100
sensory (clinical diagnosis)	6	
sensorimotor (clinical diagnosis)	3	
sensory axonal	12	
sensorimotor axonal	4	
not classified	7	
bilateral vestibulopathy	19/26	73
autonomic dysfunction	4/26	15
chronic cough	17/25	68
other non-cerebellar signs	6/26	56
reported symptoms (number of patients):	hyperkinesia (1), hyperreflexia (1), pyramidal sign (1), restless legs syndrome (1), restless legs syndrome and fasciulations (1), chorea and cognitive impairment (1)	
Combination of symptoms		
neuropathy and cerebellar ataxia	7/27	26
neuropathy, cerebellar ataxia, bilateral vestibulopathy (CANVAS triad)	20/27	74

[a] If not given individually: autonomic dysfunction, bilateral vestibulopathy (BVP), signs of the upper or lower motor neuron, movement disorder, relevant cognitive impairment.

Supplementary Table 7. Repeat configurations, frequency, pathogenicity and size range of *RFC1* repeats in 513 patients.

Repeat configuration	frequency	pathogenicity	size range
$(AAAAG)_{11}$	50.0%	non-pathogenic	11 RU
$(AAAAG)_{exp}$	29.1%	non-pathogenic	29–155 RU
$(AAGAG)_{exp}$	2.8%	likely non-pathogenic	30–70 RU
$(AAAGGG)_{complex}$[a]	2.6%	likely non-pathogenic	60–150 RU
$(AAGGG)_{<250RU}$	0.5%	non-pathogenic	83–220 RU
$(AAAGG)_{<500RU}$	1.1%	non-pathogenic	94–360 RU
$(AAGGG)_{>250RU}$[b]	10.6%	pathogenic	325–1395 RU[2]
$(AAAGG)_{>500RU}$	0.9%	pathogenic	526–775 RU
$(ACGGG)_{complex}$[c]	1.6%	unknown significance	40–80 RU
$(ACAAG)_{exp}$	0.4%	unknown significance	165–420 RU
$(AGGGG)_{exp}$	0.2%	unknown significance	50–65 RU
$(AACGG)_{exp}$	0.1%	unknown significance	80 RU
$(AAAGG)_{317}(AAGGG)_{210}$	0.1%	unknown significance	527 RU

[a] 'AAAGGG' was not observed as pure expansion, but interrupted by stretches of 'AAGGG' and 'AAAGGG'.

[b] For 40/108 alleles a large expansion (>250 RU) could be confirmed without being able to determine the exact size.

[c] Similar to $(AAAGGG)_{complex}$, 'ACGGG' was not observed as a pure expansion, but interrupted by stretches of 'AAGGG' and 'AAAGGG'.

Supplementary Table 8. Diagnoses of hereditary ataxia identified by NGS sequencing.

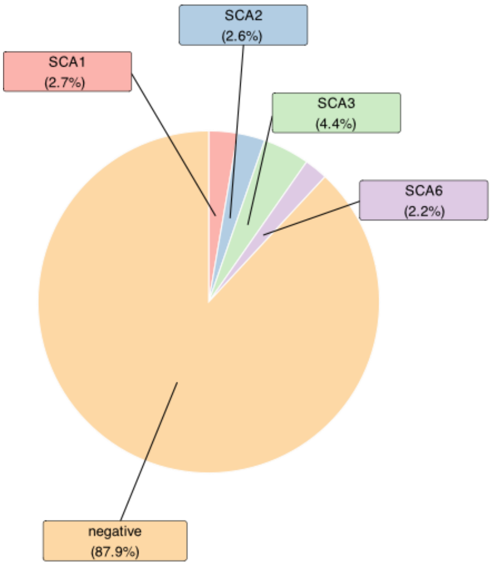
Pat.	Diagnosis	Gene	Inheri-tance	Variant(s)	Classification
4	SCA11 possible	*TTBK2*	AD	NM_173500.4: c.1319T>G p.(Val440Gly), heterozygous	Class 3 (PM2_sup, BP4)
7	SCA13	*KCNC3*	AD	NM_004977.3: c.1259G>A p.(Arg420His), heterozygous	Class 5 (PS3, PS4_mod, PP1_mod, PM2_sup, PP2)
47	SPG39 possible	*PNPLA6*	AR	NM_001166114.2: c.4051C>T p.(Arg1351*), heterozygous; NM_001166114.2: c.832G>A p.(Ala278Thr), heterozygous	class 4 (PVS1_mod, PS4_mod, PM3, PM2_sup, PP1); class 3 (PM2_sup, PP2)
55	SCA11	*TTBK2*	AD	NM_173500.4: c.1306_1307del p.(Asp436Tyrfs*14), heterozygous	Class 5 (PVS1, PS4_mod, PM2_sup)
62	EA2 possible	*CACNA1A*	AD	NM_001127222.2: c.2069C>T p.(Ser690Phe) heterozygous	Class 3 (PP3_mod, PM2_sup, PP2)
84	POLR3A-associated ataxia	*POLR3A*	AR	NM_007055.4c.1771-7C>G p.?, homozygous	Class 4, PM3, PP1_mod, PS3_sup, PS4_sup, PM2_sup
85	SCAR8	*SYNE1*	AR	NM_182961.4: c.10414C>T p.(Arg3472*), heterozygous; NM_182961.4: c.18816-50_23302-551dup (exons 101-128) heterozygous	Class 5 (PVS1, PM2_sup, PM3_sup); Class 4 (PVS1_str, PM2_sup, PM3)
110	SCA42 possible	*CACNA1G*	AD	NM_018896.5: c.769C>T p.(Arg257Cys), heterozygous	Class 3 (PM2_sup, PP2)
127	SPG79A	*UCHL1*	AD	NM_004181.5: c.333del p.(Ser112Glnfs*3), heterozygous	Class 4 (PVS1, PM2_sup)
138	ataxia telangiectasia	*ATM*	AR	NM_000051.4: c.6188G>A p.(Gly2063Glu), homozygous	Class 4 (PS3_mod, PM3, PS4_sup, PM2_sup)
143	Developmental and epileptic encephalopathy	*STXBP1*	AD	NM_001032221.6: c.246+1G>A p.?, mosaicism (VAF ca. 29%)	Class 5 (PVS1, PS4_sup, PM2_sup)
151	EA2	*CACNA1A*	AD	NM_001127222.2: c.4478A>G p.(Tyr1493Cys), heterozygous	Class 4 (PM1, PP3_mod, PM2_sup, PP2)
166	SCA28 possible	*AFG3L2*	AD	NM_006796.3: c.1998G>A p.(Met666Ile), heterozygous	Class 3 (PM5, PP3_mod, PM2_sup)
202	SPG7[a]	*SPG7*	AR	NM_003119.4: c.739C>T p.(Arg247*), heterozygous; NM_003119.4: c.1529C>T p.(Ala510Val), heterozygous	Class 5 (PVS1, PS4_mod, PM3, PM2_sup); Class 5 (PM3_str, PS4_mod, PP3_mod, PS3_sup, PP1)
233	SCAR10	*ANO10*	AR	NM_003119.4: c.1150_1151del p.(Leu384Asnfs*91), homozygous	Class 5 (PVS1, PM3, PS4_sup, PM2_sup)
257	NARP syndrome	*MT-ATP6*	mitochondrial	ENST00000361899.2: c.665T>C p.(Leu222Pro), heteroplasmic (VAF 95%)	Class 4
258	SCA48 possible[b]	*STUB1*	AD	NM_005861.4: c.799T>C p.(Phe267Leu) heterozygous	Class 3 (PM2_sup)
261	SCA14 possible	*PRKCG*		c.173G>A p.(Gly58Arg), heterozygous	Class 3 (PP3_mod, PM2_sup, PP2)
313	SCA42	*CACNA1G*	AD	NM_018896.5: c.5144G>A p.(Arg1715His), heterozygous	Class 5 (PP3_str, PS3_mod, PS4_mod, PM2_sup, PP1, PP2)
317	SPG5A[c]	*CYP7B1*	AR	NM_004820.5: c.1456C>T p.(Arg486Cys), homozygous	Class 5 (PM3_str, PP1, PP3, PP4))

375	SCA42 possible	*CACNA1G*	AD	NM_018896.5: c.2081A>G p.(Tyr694Cys), heterozygous	Class 3 (PP3_mod, PM2_sup, PP2)
393	Tay-Sachs disease possible	*HEXA*	AR	NM_000520.6: c.805G>A p.(Gly269Ser), heterozygous; NM_000520.6: c.1074-7_1074-3 del p.?, heterozygous	Class 5 (PM3_str, PS4_mod, PM5, PP3_mod, PS3_sup, PM2_sup); Class 3 (PM3, PM2_sup, PP1, PP3)
394	SPG7	*SPG7*	AR	NM_003119.4: c.1529C>T (p.Ala510Val), homozygous	Class 5 (PM3_str, PS4_mod, PM3_mod, PS3_sup, PP1)

[a] additionally a repeat expansion in *FGF14* with currently unknown pathogenicity (180–249 RU) was identified

[b] additionally reduced-penetrance expansion in *TBP* was detected; possible diagnoses: SCA17, SCA48, SCA48 modified by a reduced-penetrance expansion in *TBP*

[c] additionally a reduced-penetrance expansion in *FGF14* was detected compatible with the diagnosis of SCA27B



Supplementary Figure 2. Frequency of SCA1–3 and 6 in PCR-based repeat analyses of patients with adult-onset ataxia within the last two decades.

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