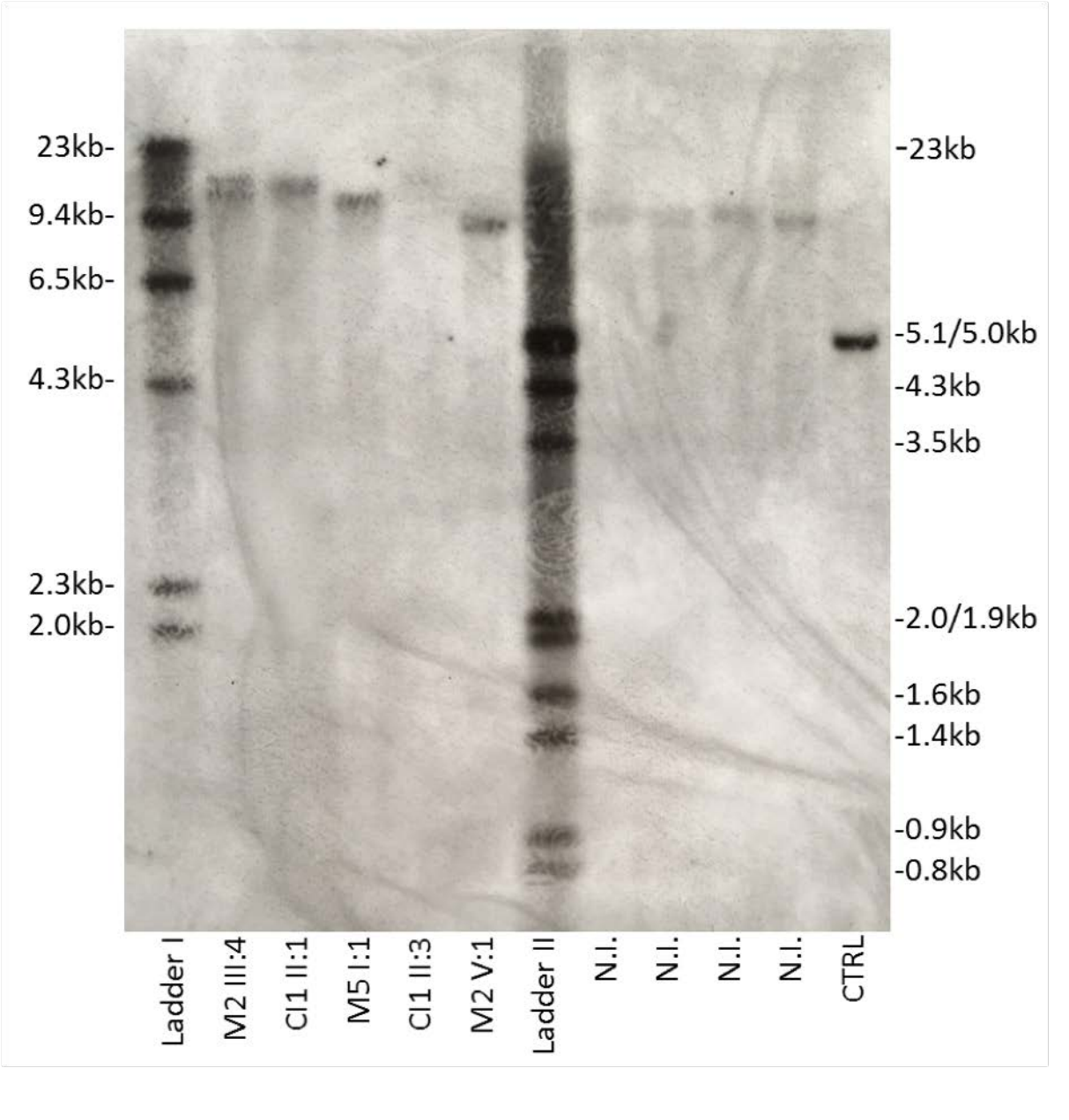
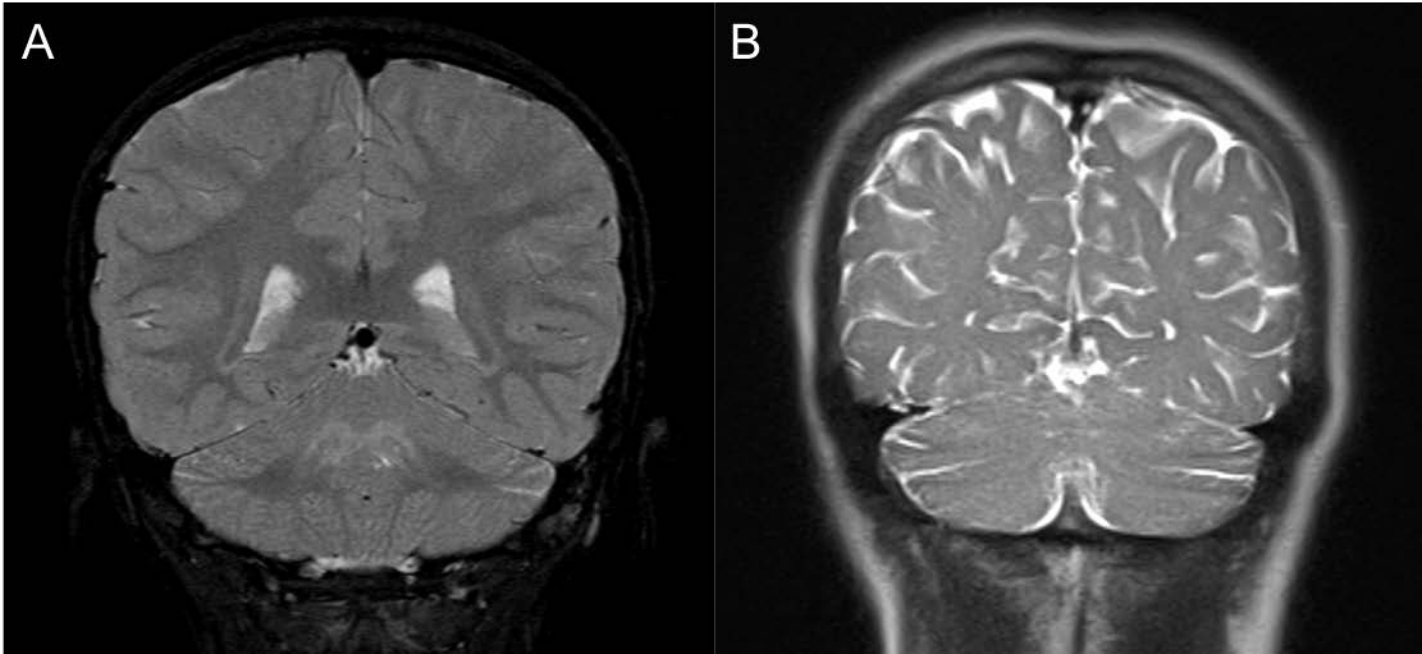
Supplementary Figure 1. Southern blotting of genomic DNA from 5 affected individuals from 2 families and 1 sporadic case. Patients show two discrete or overlapping bands of 8.9 to 13.7 kb. The sample from CI1 II:3 failed due to low DNA quality or quantity. In the control sample (CTRL), one 5-kb band corresponding to the expected size for reference allele $(AAAAG)_{11}$ is observed. Ladders used are DIG-labelled DNA Molecular Weight Marker II (Roche) (LADDER I, left) containing 8 fragments with the following base pair lengths: 125 (not shown), 564 (not shown), 2027, 2322, 4361, 6557, 9416, and 23,130 bp and DIG-labelled DNA Molecular Weight Marker III (Roche) (LADDER II, right) containing 13 fragments with the following base pair lengths: 125 (not shown), 564 (not shown), 831, 947, 1375, 1584, 1904, 2027, 3530, 4268, 4973, 5148, and 21,226 bp. N.I., sample not included in this study.


23kb-
9.4kb-
6.5kb-
4.3kb-
2.3kb-
2.0kb-
-23kb
-5.1/5.0kb
-4.3kb
-3.5kb
-2.0/1.9kb
-1.6kb
-1.4kb
-0.9kb
-0.8kb
Ladder I
M2 III:4
CI1 II:1
M5 I:1
CI1 II:3
M2 V:1
Ladder II
N.I.
N.I.
N.I.
N.I.
CTRL

Supplementary Figure 2. MRI scans of individual M2 V:1. A. Age 6: Coronal T2 showing preservation of cerebellar mass but with abnormal signal in the superior deep cerebellar white matter which extended into the superior cerebellar peduncles. B. Age 15: Haste Coronal T2 scan showing mild generalised supratentorial and cerebellar volume loss but with particular prominence of the horizontal cerebellar fissure indicating crus 1 atrophy.


A
B

Supplementary Table 1. List of genes on the broad neurogenetic gene panel used for patient M2 V:1.

Genes on broad neurogenetic panel
AARS
ABCB7
ABCD1
ABHD12
ACTB
ACTG1
ADAR
ADCK3
ADGRG1
AFG3L2
AIFM1
AIMP1
AKT1
AKT3
ALDH18A1
ALDH3A2
ALS2
AMPD2
ANG
ANO10
ANO3
AP4B1
AP4E1
AP4M1
AP4S1
AP5Z1
APTX
ARFGEF2
ARHGEF10
ARL6IP1
ARSA
ARSI
ARX
ASAH1
ASCC1
ASPA
ASPM
ATCAY
ATL1
ATL3
ATM
ATP1A3
ATP2B3

ATP2B4
ATP6AP2
ATP7A
ATR
B3GALNT2
B4GALNT1
BCAP31
BEAN1
BICD2
BSCL2
C12orf65
C19orf12
CACNA1A
CACNA1B
CACNA1G
CACNB4
CCT5
CDK5RAP2
CENPJ
CEP152
CHCHD10
CHMP2B
CLCN2
CLPP
COASY
COL4A1
COL4A2
COX6A1
CPT1C
CSF1R
CTDP1
CUL3
CWF19L1
CYP2U1
CYP7B1
DARS2
DCAF8
DCTN1
DCX
DDHD1
DDHD2
DENR
DHTKD1
DNAJB2
DNAJB5

DNM2
DNMT1
DRD2
DRP2
DST
DYNC1H1
EGR2
EIF2B1
EIF2B2
EIF2B3
EIF2B4
EIF2B5
ELOVL4
ELOVL5
ENTPD1
EOMES
ERLIN1
ERLIN2
EXOSC3
EXOSC8
FA2H
FAM126A
FAM134B
FARS2
FAT3
FAT4
FBLN5
FBXO38
FGD4
FGF14
FIG4
FKRP
FKTN
FLNA
FLRT1
FLVCR1
FTL
FUS
FXN
GAD1
GALC
GAN
GARS
GBA2
GCH1

GDAP1
GFAP
GJB1
GJB3
GJC2
GNAL
GNB4
GOSR2
GRID2
GRM1
HACE1
HARS
HEPACAM
HINT1
HK1
HNRNPA1
HNRNPA2B1
HNRNPUL1
HNRNPUL2
HOXD10
HPCA
HSPB1
HSPB3
HSPB8
HSPD1
IBA57
IFIH1
IGHMBP2
IKBKAP
INF2
ISPD
ITPR1
KARS
KCNA1
KCNC3
KCND3
KCTD13
KIAA0196
KIF1A
KIF1B
KIF1C
KIF21A
KIF2A
KIF5A
KIF5C

KLC2
KLC4
KTN1
L1CAM
LARGE1
LAS1L
LITAF
LMNA
LMNB1
LRSAM1
LYST
MAG
MARS
MARS2
MATR3
MCPH1
MED25
MFN2
MLC1
MORC2
MPZ
MRE11A
MTMR2
MTPAP
MYH14
MYH7
NAGLU
NDE1
NDRG1
NEFL
NGF
NIPA1
NOP56
NOTCH3
NT5C2
NTRK1
OPA1
OPTN
OTUD4
PAFAH1B1
PANK2
PARK2
PAX6
PCNT
PDK3

PDYN
PEX10
PFN1
PGAP1
PIK3R5
PLA2G6
PLEKHG5
PLP1
PMP2
PMP22
PMPCA
PNKP
PNPLA6
POLG
POLR3A
POLR3B
POMGNT1
POMGNT2
POMK
POMT1
POMT2
PPP2R2B
PRDM12
PRKCG
PRKRA
PRNP
PRPH
PRPS1
PRRT2
PRX
PSAP
PTEN
RAB3GAP2
RAB7A
REEP1
REEP2
RELN
RNASEH2B
RNASET2
RNF170
RNF216
RPIA
RTN2
RTTN
RUBCN

SACS
SAMHD1
SEPT9
SBF1
SBF2
SCN11A
SCN1A
SCN9A
SCYL1
SETX
SGCE
SH3TC2
SIGMAR1
SIL1
SLC12A6
SLC16A2
SLC17A5
SLC1A3
SLC20A2
SLC25A46
SLC2A1
SLC33A1
SLC52A1
SLC52A2
SLC52A3
SLC5A7
SLC6A3
SLC9A1
SNX14
SOD1
SOX10
SPAST
SPG11
SPG20
SPG21
SPG7
SPR
SPTBN2
SPTLC1
SPTLC2
SQSTM1
STUB1
SYNE1
SYT14
TAF1

TARDBP
TBCD
TBK1
TBP
TBR1
TDP1
TECPR2
TFG
TH
THAP1
TNFAIP1
TOR1A
TOR1AIP1
TPP1
TREX1
TRIM2
TRIP4
TRPV4
TSEN2
TSEN34
TSEN54
TTBK2
TTPA
TTR
TUBA1A
TUBB
TUBB2B
TUBB3
TUBB4A
TUBG1
UBA1
UBA5
UBQLN2
UNC79
UNC80
USP8
VAMP1
VAPB
VCP
VLDLR
VPS13A
VPS37A
VRK1
WDR45
WDR48

WDR62
WNK1
WWOX
YARS
YWHAE
ZBTB18
ZFR
ZFYVE26
ZFYVE27

Supplementary Table 2. Key clinical information for affected patients. d. Deceased; ↓: Decreased/reduced.↑: Increased. AJ: Ankle jerks. Av: Average. BVL: Bilateral vestibular loss. CMAP: Compound muscle action potential. DBN: Downbeat nystagmus. Hor: Horizontal. LL: Lower limb. N: normal. N/A: not assessed. NCS: Nerve conduction studies. ND: not documented. Onset: age at symptom onset. SNAPs: Sensory nerve action potentials. TG: Tandem gait. UL: Upper limb. Vert: Vertical. vHIT: Video head impulse test. VVOR: Video visually enhanced vestibulo-ocular reflex. *Patient assessed early in disease course due to having affected sibling known to Neurology service.

Patient details	Pertinent findings from first neurological examination							Progression	Investigations		
Patient Sex Onset	**Gait**	**Dysarthria**	**Limb ataxia**	**Eye movements & head impulse**	**Reflexes**	**Sensation**	**Other**	**Years from onset to disability milestone**	**Brain MRI**	**NCS**	**VHIT**
C1 II:1 F 48	Ataxic	Mild	Bilateral	Saccadic breakdown of pursuit. Ocular dysmetria	N	N	Syncope	Walker 5; Wheelchair 18	Brainstem & cerebellum atrophy 50 yrs	N/A	BVL. Av gain R0.14, L0.15. 63 yrs
C1 II:3 M 45	Ataxic	Severe	Bilateral	Square wave jerks. Broken pursuit	Absent AJ	N	Dizziness, urinary urgency, chronic cough	Walker 10	Cerebellar vermian & hemispheric parenchymal atrophy 51 yrs	↓ SNAPs. Normal motor study	BVL. Av gain R0.23, L0.33. 53 yrs
M2 III:1 M 67	Impaired TG	N	N	N	Absent AJ	↓ foot pinprick	Chronic cough	Independent	N/A	N/A	N/A
M2 III:2 F 49	Ataxic	Severe	UL & LL	Nystagmus on hor. gaze to the right	↓ AJ	ND	Syncope, urinary incontinence	Walker 6; Wheelchair 9; Bedbound 11	N/A	N/A	N/A
M2 III:3 M 57	Impaired TG	Moderate	Mild UL	N	↓ UL & LL	↓ knee vibration sense	Foot pain, chronic cough	Walker 8	Normal 63 yrs	↓ SNAPs. Mildly ↓ LL motor conduction velocities.	BVL. Av gain R0.33, L0.31. Abnormal VVOR 64 yrs
M2 III:4 F 46	Impaired TG	Mild	N	N	Absent AJ	N	Chronic cough	Stick 10; Walker 15	Normal 51 yrs, cerebellar atrophy 62 yrs	Absent SNAPs. ↓ CMAP amplitudes	N/A
M2 III:5 M 59	ND	ND	ND	ND	ND	ND	Hand/foot pain, chronic cough	Independent	N/A	N/A	Catch up saccades. Av gain R 0.67, L0.75. 58 yrs
M2 V:1 F 6	Impaired TG	Moderate	Bilateral	N	Upgoing plantars	N	Numb feet	Wheelchair 2	Dorsal brainstem & cerebellar peduncle T2 hyperintensity 7 yrs	N/A	BVL. Av gain R0.46, L.49. 18 yrs
M3 I:1 M 84	Ataxic	Moderate	Bilateral	DBN on hor. gaze in both directions	ND	↓ knee vibration sense, ↓ toe proprioception	Numb feet, chronic cough	Walker < 1 yr	Cerebellar atrophy 84 yrs	↓ SNAPs. Normal motor study.	BVL. Av gain R0.23, L0.28. 88 yrs
M4 I:1 F 53	Impaired TG	N	Mild UL	DBN in primary position. Abnormal head impulse	N	↓ pinprick & vibration below ankles	Numbness, burning pain	Independent	Mild ventral vermian & Crus I atrophy 54 yrs	↓ SNAPs. Normal motor study.	BVL. Av gain R-0.04, L-0.03. 62 yrs
M5 I:1 M 50	Ataxic	Moderate	N	Rotatory & DBN in all directions. Occasional square wave jerks	↓ UL	↓ vibration LL, ↓ left toe joint position sense	Burning pain in feet, chronic cough	Walker 15	Moderate dorsal & severe ventral vermian atrophy 73 yrs	Absent SNAPs ↓ CMAP amplitudes. Mildly ↓ motor conduction velocities	BVL. Av gain R0.06 L0.04. 77 yrs
M6 I:1 F 54	Ataxic	Mild	UL	DBN on hor. gaze	N	↓ whole limb pinprick, ↓ big toe proprioception	Chronic cough	Walker 4	Cerebellar & Crus I atrophy 56 yrs	↓ SNAPs. Normal motor study	BVL. Av gain R0.4, L0.51. Abnormal VVOR. 51 yrs
M7 I:1 M 56	Impaired TG	Mild	Bilateral	DBN. Positive head impulse	↑ at triceps, ↓ AJ	↓ whole limb vibration	Postural dizziness	Crutches 2; Walker 3	Mild ventral vermian & Crus I atrophy 56 yrs	Absent SNAPs. Normal motor study	BVL. Av gain R0.22, L0.21. 60 yrs

Marker ID	Position	Patient ID				Annotation
		C1 II:1	C1 II:3	M2 III:4	M2 V:1	
rs6814900	chr4:38157510	1/2	1/2	1/1	1/1	
rs2011590	chr4:38944101	1/1	1/1	1/1	1/1	Cohort specific haplotype
rs7694333	chr4:38952030	1/1	1/1	1/1	1/1	Cohort specific haplotype
rs17583377	chr4:38955135	1/1	1/1	1/1	1/1	Cohort specific haplotype
rs6841592	chr4:38965986	1/1	1/1	1/1	1/1	Cohort specific haplotype
rs1864495	chr4:39036311	1/1	1/1	1/1	1/1	Cohort specific haplotype
rs2566120	chr4:39055870	1/1	1/1	1/1	1/2	Cohort specific haplotype
rs2566134	chr4:39074295	2/2	2/2	2/2	2/2	Same extended haplotype as Cortese *et al.*
rs3733275	chr4:39122697	1/1	1/1	1/1	1/1	Same extended haplotype as Cortese *et al.*
rs2044917	chr4:39149491	1/1	1/1	1/1	1/1	Same extended haplotype as Cortese *et al.*
rs2711991	chr4:39151353	1/1	1/1	1/1	1/1	Same extended haplotype as Cortese *et al.*
rs2062229	chr4:39286766	2/2	2/2	2/2	2/2	Same extended haplotype as Cortese *et al.*
rs2066790	chr4:39318706	1/1	1/1	1/1	1/1	Same core haplotype as Cortese *et al.*
rs11096992	chr4:39329102	2/2	2/2	2/2	2/2	Same core haplotype as Cortese *et al.*
rs17584703	chr4:39364856	2/2	2/2	2/2	2/2	Same core haplotype as Cortese *et al.*
rs6844176	chr4:39366590	1/1	1/1	1/1	1/1	Same core haplotype as Cortese *et al.*
rs6823497	chr4:39385821	1/1	1/1	1/1	1/1	Cohort specific haplotype
rs13135439	chr4:39405151	2/2	2/2	2/2	2/2	Cohort specific haplotype
rs11940694	chr4:39414993	2/2	2/2	2/2	2/2	Cohort specific haplotype
rs7674434	chr4:39419409	1/1	1/1	1/1	1/1	Cohort specific haplotype
rs13129975	chr4:39526641	2/2	2/2	1/1	1/1	
rs2381387	chr4:39531928	2/2	2/2	2/2	2/2	
rs10026108	chr4:40379061	2/2	2/2	2/2	2/2	
rs10008483	chr4:40712481	2/2	2/2	2/2	2/2	

Supplementary Table 3. Comparison of core haplotype identified from Cortese *et al.* (2019) with patients from this cohort with WGS sequencing data. Marker SNPs are the same as Cortese et al. (2019). Our patients share the 'core' haplotype region identified in Cortese et al. (2019). Beyond that is an additional shared region that is specific to our cohort, suggesting a more recent common ancestor for our patients.

Marker ID	Position	C1 II:1	C1 II:3	M2 III:4	M2 V:1	M3 I:1	M4 I:1	M5 I:1	M6 I:1	M7 I:1
rs9995922	chr4:36316348	1/2	1/2	1/1	1/1	1/1	1/1	1/1	1/2	1/1
rs10049643	chr4:36815755	1/1	1/1	1/1	1/1	1/1	1/1	1/1	1/1	1/1
rs2973217	chr4:37430794	1/1	1/1	1/1	1/1	1/2	1/1	1/2	1/2	1/2
rs2276943	chr4:37635128	1/2	1/2	2/2	2/2	2/2	2/2	1/2	2/2	1/1
rs6531590	chr4:37855485	1/1	1/1	1/1	1/1	0/0	0/0	0/0	0/0	0/0
rs2279027	chr4:37902135	1/1	1/1	1/1	1/1	1/1	1/1	1/2	1/2	1/2
rs623945	chr4:38118264	2/2	2/2	2/2	2/2	1/1	2/2	2/2	2/2	2/2
rs2637706	chr4:38510250	2/2	2/2	2/2	2/2	0/0	0/0	2/2	0/0	0/0
rs17616226	chr4:38689805	2/2	2/2	2/2	2/2	2/2	2/2	2/2	2/2	2/2
rs5743810	chr4:38828729	2/2	2/2	2/2	2/2	2/2	2/2	2/2	2/2	2/2
rs2271031	chr4:38935751	2/2	2/2	2/2	2/2	1/2	2/2	2/2	2/2	2/2
rs3733287	**chr4:39215732**	1/1	1/1	1/1	1/1	1/1	0/0	1/1	0/0	1/1
rs4975020	chr4:39527132	1/1	1/1	1/1	1/1	0/0	1/1	1/1	1/1	1/1
rs7688174	chr4:40243362	1/1	1/1	1/1	1/1	0/0	0/0	1/1	0/0	0/0
rs10022491	chr4:40335891	1/2	1/2	2/2	1/2	1/1	2/2	2/2	2/2	2/2
rs278981	chr4:40425993	1/1	1/1	1/1	1/1	1/2	1/1	1/1	1/1	1/1
rs278950	chr4:40502030	1/1	1/1	1/1	1/1	1/1	1/1	1/1	1/1	1/1
rs4861066	chr4:40808646	1/1	1/1	1/1	1/1	1/2	1/1	1/1	1/1	1/1
rs10025369	chr4:41014467	2/2	2/2	1/2	1/1	0/0	0/0	1/2	2/2	0/0
rs17528160	chr4:41263904	1/1	1/1	1/1	1/1	1/1	0/0	1/1	0/0	0/0
rs4266323	chr4:41524486	0/0	1/1	1/1	2/2	1/2	2/2	2/2	2/2	2/2
rs17528897	chr4:41661604	2/2	2/2	2/2	1/1	1/2	2/2	1/2	2/2	1/2
rs12643893	chr4:42067012	1/1	1/1	1/1	1/1	1/1	1/1	1/1	1/1	1/2
rs898500	chr4:42637169	1/1	1/1	1/2	1/1	1/2	1/1	1/1	1/1	1/2
rs4861050	chr4:43030595	1/1	1/1	1/1	1/1	1/1	1/1	1/1	0/0	1/1
rs10022054	chr4:43899131	1/2	1/2	1/1	1/1	1/1	1/1	1/2	0/0	1/1
rs13117500	chr4:44293822	2/2	2/2	2/2	1/2	2/2	2/2	2/2	2/2	2/2

Supplementary Table 4. Haplotyping summary across all patients with next-generation sequencing data. Highlighted regions are homozygous shared haplotypes.