Supplementary Note 1. Summary of clinical findings in the reported families

Family 1. *NAA60,* c.321_327del (p.Arg108Thrfs*3)

In Family 1, two siblings from a non-consanguineous British family were included in this study. The proband, case F1-II-1 had a normal birth and normal development until the early twenties. She presented with a combination of movement, cognitive and psychiatric features. She first noticed episodes of unsteadiness; brief loss of consciousness followed by falls when she was in her twenties. An electroencephalogram (EEG) performed at the time did not reveal the cause of these events. At the age of 42 years, she presented to neurology as the falls had increased in frequency. She started falling backwards and her gait became broad-based and was unstable when turning around corners. Her brain CT scan revealed extensive symmetrical calcifications involving basal ganglia, the pons, and the cerebellar structures. Investigations excluded any acquired and metabolic causes of brain calcifications. Her condition progressed and five years into the disease the ataxia-related symptoms deteriorated, her speech became dysarthric and she started to experience word-finding difficulties with significant deterioration in her memory. She developed severe depression requiring medication. Her neurological examination showed a combination of cerebellar ataxia with new additional features of parkinsonism (bradykinesia and rigidity) and cognitive impairment, being unable to follow simple instructions. Neuropsychology testing revealed functioning in the impaired range on an un-timed measure of non-verbal abstract reasoning, suggesting significant decline in non-verbal intellectual abilities. Focal testing revealed grossly impaired attentional and executive functioning and slowed speed of processing. Visual perception was also impaired. Performance was poor on measures of language comprehension and recognition memory. Overall, the profile suggested profound cognitive dysfunction, which although mostly implicated anterior and subcortical functions, was global in nature.

As the condition continued to progress, neurological examination 10 years into her disease, showed significant deterioration. She became wheelchair bound, requiring help with

most daily activities, and developed severe headaches. Neurological examination revealed a combination of severe bradykinesia, rigidity, dystonic postures in the neck and limbs, broken pursuit, reduced up-gaze, dysarthria, weak cough with poorly protruding tongue. Levodopa treatment had a modest benefit at the onset of the disease, but limited benefit was recorded at this stage. On the last examination, 17 years into the disease, she presented with a combination of cognitive deficit, severe extrapyramidal and cerebellar syndrome, severe speech and swallowing difficulties, becoming anarthric and unable to swallow, requiring percutaneous endoscopic gastrostomy (PEG) feeding. At this stage, there were extensive brain calcifications on brain imaging.

Her younger brother, case F1-II-2, had a normal development and health until his twenties. His symptom at onset was severe psychosis requiring hospitalization. He continued to have visual and auditory hallucinations and was subsequently diagnosed with Schizophrenia. Despite medical treatment, the hallucinations were refractory to treatment. A CT scan performed at the age of 39 showed extensive symmetrical brain calcifications involving bilateral basal ganglia. Despite a different onset, the disease progressed in a similar way to his sibling. He started having walking difficulties with a combination of cerebellar and parkinsonian syndrome. Ten years from onset of psychiatric symptoms he needed walking aids and three years after that, he became bed bound. On last examination, 15 years from onset of the disease, the patient required full-time care, was anarthric, unable to swallow and being PEG-fed, presenting very limited movement due to severe rigidity and bradykinesia.

Family 2. *NAA60,* c.338-1G>C, p.(Gly113Valfs*32)

Family 2 includes two siblings from a consanguineous family of Algerian descent. The proband had normal development until her early thirties. She worked as a restaurant manager. At the age of thirty she presented generalized seizures which led to the discovery of symmetric and extensive brain calcifications involving basal ganglia, cerebellar hemispheres, and fronto-parieto-temporo-occipital cortex. Despite these widespread calcifications, neurological examination was normal. There was no cerebellar ataxia. She could stand more than 10s on

monopodial support and walk a tightrope. She had neither pyramidal tract irritation signs nor extrapyramidal symptoms. EEG was normal. Acquired and metabolic causes of brain calcifications were excluded, as well as variants of *POLG, MELAS, MERRF, SLC20A2, PDGFB, PDGFRB, XPR1 and TREX1.* CSF study and eye examination were normal. During follow up, she presented insular and temporal seizures easily controlled with lamotrigine. Though she did not develop cerebellar, extrapyramidal, or sensorimotor symptoms, she complained of progressive memory and impulse control disorders. Neuropsychological examination revealed frontal with attention deficit and dysexecutive syndrome, and visuospatial impairment. Two years later, she insidiously developed slight bradykinesia without abnormal gait, which did not require dopamine medication nor physiotherapy. Five years after the initial diagnosis, she is still under lamotrigine monotherapy and does not present significant disability except frontal impairment.

Since childhood, her brother presented an intellectual disability with a delay in speech development since childhood. Nevertheless, he followed a normal schooling until the age of 14. Afterward, he worked as a mechanic in a centre for people with disabilities. In his late twenties he developed a progressive cerebellar syndrome, seizures and extrapyramidal signs which required physiotherapy, lamotrigine, and dopamine medication. A CT-scan performed at the age of 43 showed extensive symmetrical brain calcifications similar to those of his sister. After four years of disease, there is no significant worsening of the clinical signs. Both siblings had macrocrania with an oblong face.

Family 3. *NAA60*, c.338-1G>C, p.(Gly113Valfs*32)

Two sisters were affected by extensive brain calcifications. They were born from consanguineous parents and originated from Morocco. The older sister, suffered from migraine without aura. Upon neurological examination at the age of 31, bilateral Babinski sign was noticed along with mild akineto-hypertonic syndrome, slight dysmetria in the context of unsteadiness, suggesting mild cerebellar syndrome. SARA scale scored 16/40 and UPDRS-III 10/108. CT-scan revealed extensive calcifications (Total Calcification Score = 46).

Metabolic screen revealed no calcium-phosphate metabolism disorder. In addition, she had a history of intellectual disability. Intellectual quotient was not assessed. Minimental state evaluation scored 18/30 at age 33. Physical examination revealed macrocrania, long face, agenesis of second, third and fourth right toes and hypoplasia of second, third and fourth left toes as well as hypopigmented regions on both upper limbs.

Her sibling was referred to an expert neurological center at the age of 24 years for the etiological assessment of extensive brain calcifications identified in the context of cephalalgia. A diagnosis of migraine without aura was performed. She complained of sadness and anxiety and of unsteadiness and fatigue. At ages 24 and 25, neurological examination was normal. However, at the age of 31, neurological examination showed a right Babinski sign and upper limb hypermetria with unsteadiness, indicating a cerebellar involvement. CT-scan revealed extensive calcifications (Total Calcification Score = 50). Metabolic screen revealed no calcium-phosphate metabolism disorder and normal blood lactate and pyruvate levels. Electro-encephalography was normal. In addition, she had a history of mild intellectual disability. Neuropsychological assessment revealed a total Intellectual Quotient of 63 (verbal IQ=63, performance IQ=64). She presented some dysmorphic features, including low-set ears and high arched palate. She also presented with coxa valga and underwent surgery in childhood for aortic coarctation and suffered from hearing impairment.

Both parents were reported to be unaffected, but could not be examined.

Family 4. *NAA60*, c.391C>T, p.His131Tyr

This family of Gujarati origin have two children and 5 miscarriages. During the pregnancy the baby was noted to be small for gestational age. There was oligohydramnios and she was born by caesarean section at 37 weeks due to breech presentation weighing 2kg. The proband had congenital cataracts, early feeding difficulties, constipation, glue ears and hyperacusis. She was referred to genetics age 10 years with ADHD (diagnosed at age 6 years), short stature (<9th centile), learning difficulties, delayed speech and language development, and dyspraxia. She exhibited sensory seeking behaviour. On examination she licked her hands repeatedly,

presented poor social skills and struggled to hold a conversation. When concentrating on tasks, she made tongue movements and had abnormal posturing. She also had low tone, sweaty palms, hypopigmented linear birthmark on her right upper arm, almond shapes eyes, and low set ears. She presented dystonia and chorea. Parents observed that she had difficulty making friends and engaging in conversations. When concentrating on tasks she would protrude her tongue and have posturing. She was treated with methylphenidate for her ADHD and had a positive response on the medication.

Her brain MRI showed symmetrical low signal in globus pallidus bilaterally and supratentorially some scattered dots and small patches of T2/FLAIR hyperintensity predominantly in the frontoparietal subcortical white matter bilaterally. She also presents generalised pruritis, multiple allergies, and congenital cataracts.

Family 5. *NAA60* c.130C>T, p.Arg44Cys

The proband was born to consanguineous parents and presented delayed motor milestones, pyramidal signs, seizures, and dystonia. He also had an abnormal EEG and dysmorphic features. Antiepileptic treatment was started due to seizures. He used multiple antiepileptics over the years due to ongoing seizures and he still does. At the last physical examination, he was microcephalic, had impaired motor development, no head control and no unassisted sitting. There were joint deformities with spasticity in the extremities. He speaks in single words, unable to form sentences. The brain MRI showed multiple T2 and Flair intense foci in the subcortical and cortical regions of the bilateral cerebral hemispheres, atrophy in the left cerebral peduncle, and cortical thickening areas compatible with polymicrogyria in the bilateral perisivian regions, temporal and parietal areas of the left cerebral hemisphere. There was no calcification in brain CT.

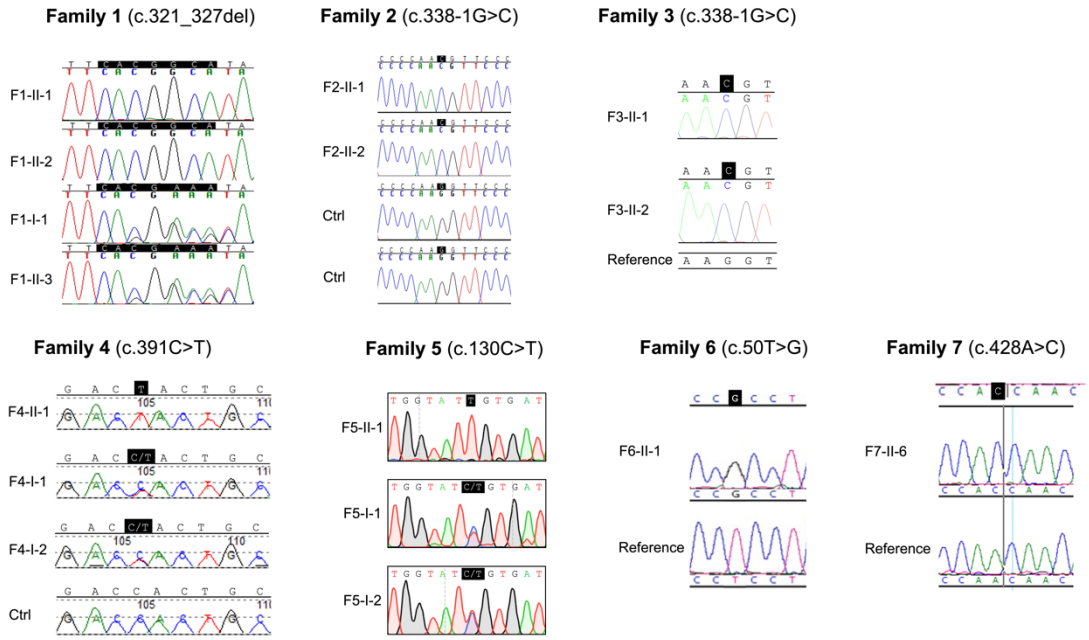
Family 6. *NAA60 c.50T>G, p.Leu17Arg*

The proband is a male from consanguineous parents from Turkey. He presented at 12 years old with motor and cognitive developmental delay, first observed at 6 months old. He has a history of seizures without fever at the age of 8-9 months and was since treated with sodium valproate. Due to hyperactivity, a child psychiatrist was consulted when he was 2 years old, and methyl phenidate was started. At 3 years old, a shift in the eyes was noticed and glasses were given due to severe astigmatism and hyperopia. At last examination, he could speak but was not clearly understood. On examination, he presents with mild intellectual disability, microcephaly, high palate, strabismus, spasticity, pyramidal syndrome (brisk reflexes, upgoing plantars), Pes planus, assisted in long-distance walking.

Family 7. *NAA60* c.428A>C, p.Asn143Thr

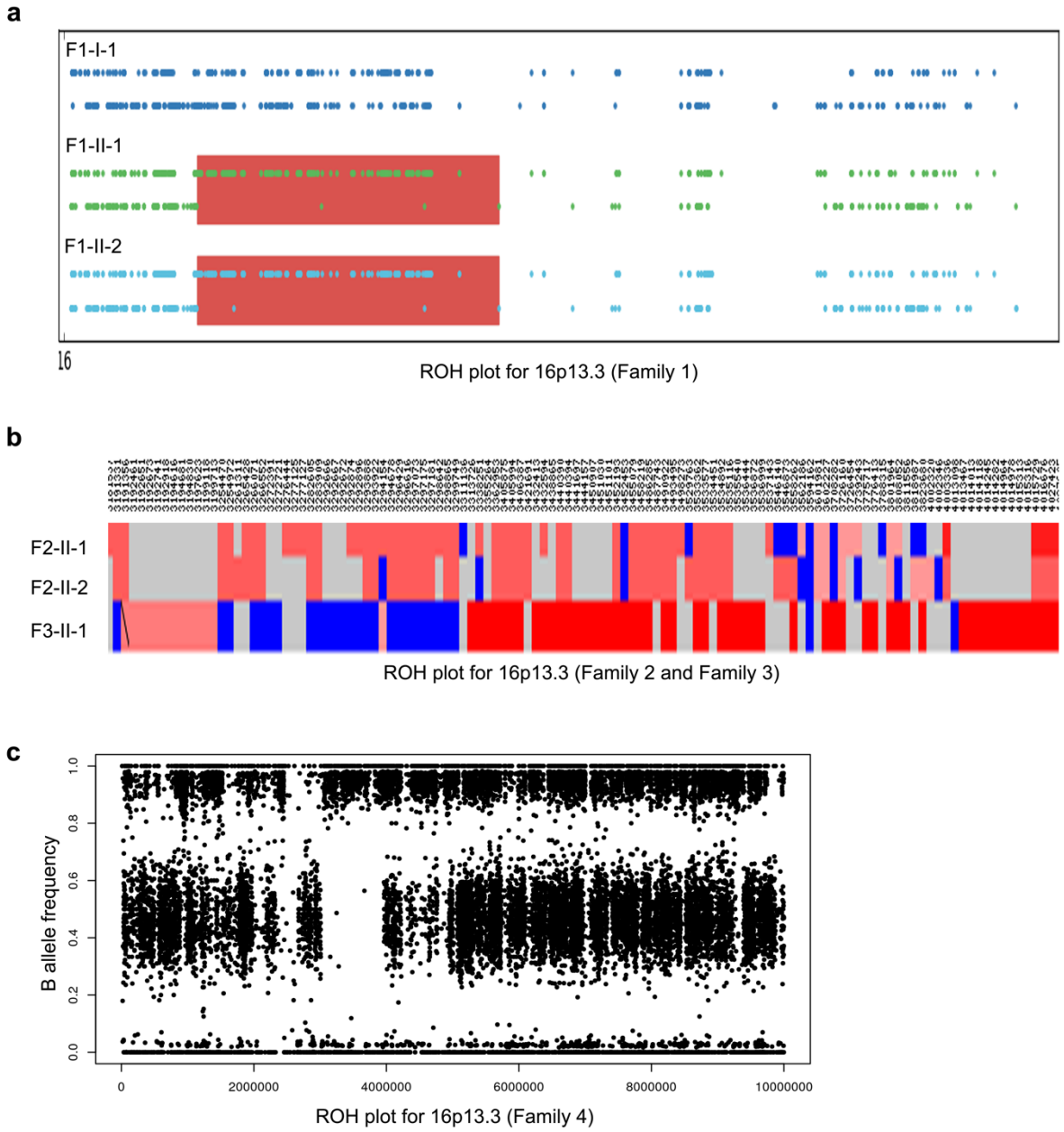
A single affected Saudi male born to non-consanguineous parents, both from the same tribe is the only affected member in kindred of nine siblings. The index had normal development until the age of 27 years when he presented with upper respiratory tract infection (high-grade fever and cough) and later developed facial asymmetry with slurred speech and decreased concentration. Past medical history included two head traumas, at 1 year old after a fall without any complication and at 7 years old after an accidental head injury that required suturing. His physical examination revealed mild cognitive impairment with MoCA score of 22/30. Neurological examination showed a combination of pyramidal and cerebellar syndrome. Brain MRI revealed multifocal ischemic changes including chronic and subacute ischemic changes, and extensive intracranial calcifications. There was also evidence of multifocal narrowing of the distal intracranial arteries compatible with intracranial vasculopathy. On CT scan, multifocal scattered intracranial coarse calcification was seen. In addition, PET-CT brain with dopa challenge of the patient gave a very unexpected pattern where dopa uptake by the basal ganglia was normal despite extensive calcification. Other investigations showed normal transthoracic echocardiography and neurophysiology examination showed no electrophysiological evidence of peripheral neuropathy in the upper or lower extremities, and EEG was normal for awake and drowsy EEG.

Supplementary Figure 1. DNA sequencing results from *NAA60* PFBC Families 1 to 7.



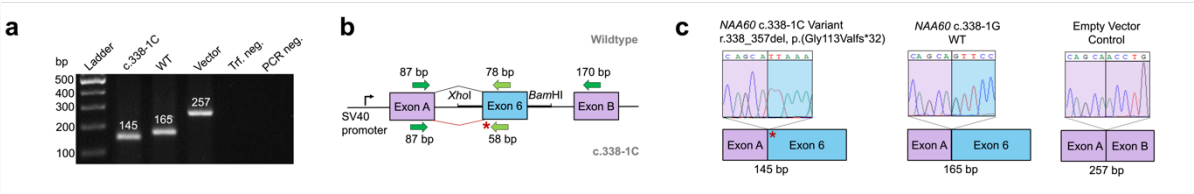
Sanger sequencing data and segregation analysis from all families. Accompanying Figure 1.

Supplementary Figure 2. Homozygosity mapping in Families 1 to 4.



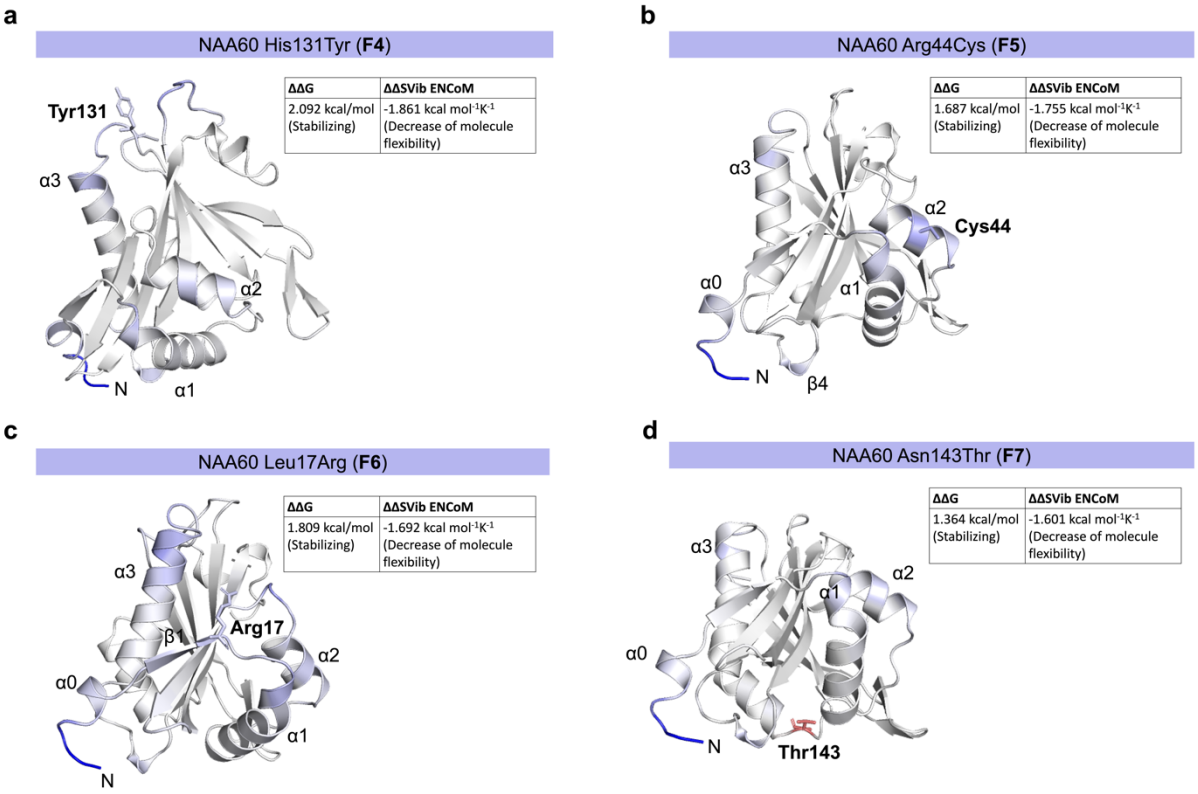
a, Three homozygous regions identified, which were shared by the two affected individuals in Family 1. **b,** regions of homozygosity (ROH) from affected members Family 2 (first two rows) and Family 3 (third row) did not show solid runs of homozygosity shared between the two families. **c,** regions of loss of heterozygosity in Family 4. These regions were on chromosome 16 and included *NAA60*. Accompanying Figure 1.

Supplementary Figure 3. RNA analysis of the *NAA60* c.338-1G>C variant confirming cryptic splice site activation.



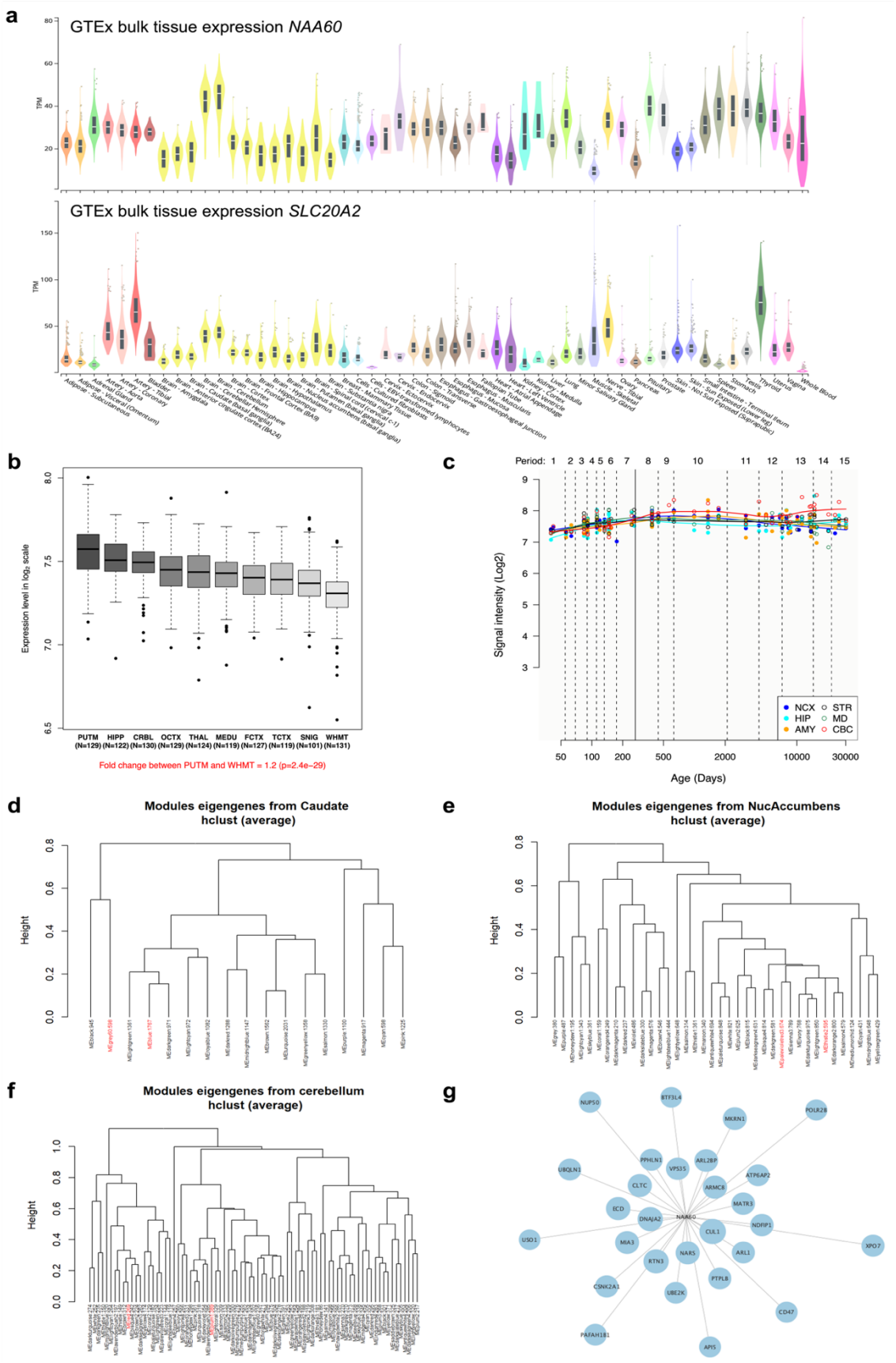
a, Gel electrophoresis of the *NAA60* c.338-1G>C variant, c.338-1G wild-type and empty pSPL3 vector amplicons. Both transfection (Trf) and PCR negative (neg) controls were performed as expected. **b,** Vector construct of the *in vitro* splice assay. Amplicons containing either the wild-type (upper splice profile in black) or variant (lower splice profile in red) were inserted between exons A and B of the pSPL3 vector. Primers used for amplification of the wild-type and variant cDNAs are shown with the dark green forward arrow annealing to Exon A (87 bp) and a light green reverse arrow annealing to exon 4 (78 bp for wild-type and 58 bp for variant). The empty vector was amplified using the two dark green arrows annealing to exons A and B. The cryptic splice activation site is depicted with an asterisk. **c,** cDNA amplicons are shown from left to right: A schematic of the c.338-1C variant with cryptic splice activation in exon 6, deleting 20 nucleotides (r.338_357del, p.(Gly113Valfs*32)) (left), wild-type c.338-1G (middle) and an empty vector control (right). Accompanying Figure 1.

Supplementary Figure 4. Predictions for differences in protein stability (ΔΔG) and flexibility (ΔΔSvib) for the missense mutants (F4-F7).



The values of folding free energy changes upon mutations (ΔΔG) were above 0 for all mutants(**a,** His131Tyr; **b,** Arg44Cys; **c,** Leu17Arg; **d,** Asn143Thr) indicating increased protein stabilization in the mutants. All mutants showed values below 0 for changes in vibrational entropy (ΔΔSVib ENCoM), indicating a loss in flexibility upon mutation. The parts of the structure with increased flexibility upon mutation are illustrated in blue. Predictions were made in DynaMut. Accompanying Figure 2.

Supplementary Figure 5. *NAA60* gene expression in various tissues.



a, GTEx gene expression of *NAA60* and *SLC20A2* and **b,** BRANIAC *NAA60* gene expression in different brain areas in adult pathologically normal human brains show that *NAA60* is expressed in all ten brain regions with highest expression detected in the putamen, cerebellum, and occipital regions (PUTM – putamen, HIPP – hippocampus, CRBL – cerebellum, OCTX – occipital cortex, THAL – thalamus, MEDU – medulla, FCTX –frontal cortex, TCTX –temporal cortex, SNIG – substantia nigra, WHMT – white matter). **c,** *NAA60* gene expression in the developing brain (NCX - neocortex; STR – striatum, HP – hippocampus, AMY – amygdala, MD – midbrain, CBC – cerebellum). **d-f,** Eigengenes correlation. In the three brain tissues of interest (cerebellum, nucleus accumbens and caudate), *NAA60*and *SLC20A2* belong to different modules within each tissue-specific network. However, for the nucleus accumbens network, these two modules are very close, and these can be visualised in the corresponding dendrogram. In each dendrogram, the modules of interest are highlighted in red. **g,** Top-Down plot of the *NAA60* module genes in the caudate tissue. Only the most connected genes are shown. Size of gene nodes reflect their connectivity with the rest of genes in the module. Proximity of genes in the plot reflects their similarity in terms of shared connections with other genes.

Supplementary Table 1. *In silico* prediction of reported NAA60 variants.

Family	**1**		**2**		**3**		**4**	**5**	**6**	**7**
Individual	F1-II-1	F1-II-2	F2-II-2	F2-II-1	F3-II-1	F3-II-2	F4-II-1	F5-II-1	F6-II-6	F7-II-6
Consanguinity	No		Yes				No	Yes	Yes	Yes
cDNA sequence	c.321_327del		c.338-1G>C				c.391C>T	c.130C>T	c.50T>G	c.428A>C
Amino acid change	(p.Arg108Thrfs*3)		p.(Gly113Valfs*32)				(p.His131Tyr)	(p.Arg44Cys)	(p.Leu17Arg)	(p.Asn143Thr)
Zygosity	Homozygous		Homozygous				Homozygous	Homozygous	Homozygous	Homozygous
AF in gnomAD	Absent		Absent				3/152156 (all South Asian)	Absent	1/17976 (East Asian) All hets	2/249122 All hets
AC in 100k GP*	1		Absent				5 (all hets)	Absent	Absent	Absent
AF in 100k GP	1/156390		0				5/156390 (All hets)	Absent	Absent	Absent
***In silico* predictions**										
SIFT	NA		NA				Deleterious (0.05)	Deleterious (0.003)	Deleterious (0)	Deleterious (0.01)
Polyphen	NA		NA				Benign (0.072)	Possibly damaging (0.855)	Probably damaging (0.95)	Probably damaging (0.99)
PROVEAN	NA		NA				Neutral (-0.87)	Damaging (-4.62)	Neutral (-2.02)	Damaging (-5.23)
Mutation Taster	Disease causing (1.0000)		Disease causing (1.0000)				Disease causing (0.9999)	Disease causing (0.9999)	Disease causing (1.0000)	Disease causing (1.0000)
CADD	NA		NA				23.9	26.4	18.5	27
Splicing Predictions	NA		MaxEnt: -100.0% NNSPLICE: -100.0% SSF: -100.0%				NA	NA	NA	NA
GERP (2.dp)	NA		NA				5.46	5.34	5.77	5.46
PrimateAI	NA		NA				Damaging (0.88)	Tolerated (0.52)	Tolerated (0.78)	Tolerated (0.426)
M-CAP	No		No				Damaging (0.6)	Damaging (0.48)	Tolerated (0.01)	Damaging (0.73)
Has been seen in other unrelated cases before	No		No				No	No	No	No
ACMG classification	Pathogenic (PVS1, PM2, PP3, PP4)		Pathogenic (PVS1, PM2, PP3, PP4)				Likely Pathogenic (PS3, PM2, PP3, PP4)	Variant of uncertain significance (PM2, PP3)	Likely Pathogenic (PM2, PP3, PS3, PP4)	Likely Pathogenic (PM2, PP3, PS3, PP4)

Abbreviations: AC = Allele count; AF = Allele frequency; 100k GP = 100 000 Genomes Project; gnomAD = The Genome Aggregation Database; hets = heterozygous); NA = not available, * N=78195 individuals in 100k GP aggregate file, ACMG=The American College of Medical Genetics and Genomics, SIFT=sorts intolerant from tolerant; PROVEAN=Protein Variation Effect Analyzer; CADD = The Combined Annotation Dependent Depletion score ; GERP= The Genomic Evolutionary Rate Profiling score; M-CAP= Mendelian Clinically Applicable Pathogenicity score.

Supplementary Note 2

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Supplementary Note 3.

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