Published by	Faber et al	Smith et al	Smith et al	Almeida et al	Kamate et al
Mutation	c.768_769dup	c.813_816del		c.900_901dup	c.912G4A
Sex	female	female	male	female	female
Age at last follow-up examination	27y (07/2018)	33y (05/2019)	36y (05/2019)	40y (03/2020)	15y (01/2020)
Parents	healthy (mother 47y, father 48y)	healthy (father 64y, mother 60y)	idem	heterozygous mother (died at 60y) FTLD + features of CBS, heterozygous father (61y) FTLDvb, severe state.	heterozygous, healthy (father 45y, mother 40y)
Other relatives	none reported ill	FTLD in maternal grandfather, paternal grandmother and great uncle	idem	maternal grandmother dementia (70y), 2 maternal siblings dementia plus parkinsonian signs (50y), 2 maternal siblings CBS (50y)	sister died at 16y, seizures (8y) and dementia, MRI cerebellar atrophy, no genetic testing
N**eurological examination**					
Motor signs	ataxia worsened, wheel-chair bound, spasticity lower extremities, increased reflexes	ataxia worsened, needs consistent help for walking	ataxia worsened, needs consistent help for walking	Early ataxia (28Y), unable to walk at 37 and at 40 y bedridden; mild pyramidal signs, dystonia	wide gait, independent in all ADL, slight gait ataxia (new)
Vision impairment	Declined, still functional	rapid vision loss (22y), almost blind (35y), retinal dystrophy	deterioration of vision (25y), almost blind, retinal dystrophy	rapid progressive visual deficit with onset at 25y; severe amaurosis at 28years, retinal dystrophy	vision still normal, fundoscop normal
Memory dysfunction	frontotemporal symptoms, severe decline, entirely dependent for ADL	no memory decline	executive functions borderline	Mild memory dysfunction (started at 36y)	borderline IQ, poor short time memory, poor analytic functions, attends school (15y), grades are falling
Psychiatric comborbidities	mood lability and delusions (onset at 23y), deteriorated to severe apathy, poor verbal contact	visual hallucinations (enhanced by AED)	mild depression	very repetitive behaviour, anxiety, emotional lability since 37y of age	none
Epilepsy features					
Seizures controlled?	no	yes	yes	yes	partially
Type(s) of seizure	Focal (photosensitive) and generalized seizures persisted.	non motor onset (visual) impaired awareness bilateral tonic clonic seizures, visual symptoms "new"	non motor onset (eye deviation) impaired awareness bilateral tonic clonic seizures	jerks elicited by sudden sounds since the beginning, but not classified as epileptic	tonic clonic seizures (1 short generalised seizure per year), photosensitivity
EEG pattern (at follow up)	slow wave background activity + epileptiform discharges in posterior regions during photostimulation	Background activity 7 Hz + brief sequences of slow waves + few spikes occipital regions	Background activity 7 Hz + brief sequences of slow waves + few spikes occipital regions	normal at beginning; follow-up EEGs refused	generalised epileptiform discharges, background activity normal (March 2019)
AEDs trialled (current AED)	LTG, LEV, CLB	barbiturates, LEV, ZNS, VPA, PER	PB, VPA, LEV, ZNS, PER	n.a.	CBZ, CLB, LEV
cMRI	global and severe cerebellar atrophy	n.a.	n.a.	severe global atrophy with marked cerebellar atrophy	cerebellar atrophy
Serum progranulin level	n.a.	<0.6ng/ml (serum)	<0.6ng/ml (serum)	<6 ng/ml (serum)	n.a.

Table 1**: Clinical follow-up data of all previously published bi-allelic GRN mutations**, FTLD: fronto-temporal lobe dementia, CBS: cortico-basal syndrome, ADL: activities of daily life, AED: anti-epileptic drug, LTG: lamotrigine, LEV: levetiracetam, CLB: clobazam, ZNS: zonisamide, VPA: valproic acid, PER: perampanel, CBZ: carbamazepine