Summary of the features of patients of HDLS

Exon	Nucleoid changes	Amino acid change	Population	Family history	Sex	Age of onset (years)	Age of death/survival time (years)	Clinical features	Brain imaging	References
2	c.49G>T	p.G17V	Chinese	No	M	26	/	Cognitive impairment Urinary incontinence Hemiplegia	Abnormal signal in lateral Ventricle Bilateral white matter lesion Thinning of corpus callosum CT: no calcifications	Wu L, 2017 (18)
/	c.310delC	p.P104LfsT*8	USA	No	F	22	/	Hemiplegia Cognitive impairment	Bilateral white matter lesion Abnormal signal in lateral ventricle Abnormal signal in frontal lobe CT: no calcifications	Miura T, 2018 (19)
12	/	p.T567fsX44	USA	NA	F	24	53/29	Hemiplegia	/	Guerreiro R, 2013 (20)
12	c.1745T>C	p.L582P	European	No	M	44	/	Gait disorder Dysphagia Psychiatric symptoms	Bilateral white matter lesion Abnormal signal in frontal lobe Thinning of corpus	Schuberth M, 2012 (21)

									callosum	
12	c.1754-2A>G	p.G585_K619delinsA	European	Yes	F	36	40/4	Cognitive impairment Psychiatric symptoms Seizures Parkinsonism	/	Rademakers R, 2012 (22)
12	c.1754-2A>G	p.G585_K619delinsA	European	Yes	F	38	41/3	Cognitive impairment Psychiatric symptoms Seizures Parkinsonism	/	Rademakers R, 2012 (22)
13	c.1765G>A	p.G589R	Japanese	No	F	44	/	Cognitive impairment Gait disorder Speech disorder Apraxia	Thinning of corpus callosum Brain atrophy Calcifications in the white matter	Daida K, 2017 (23)
13	c.1765G>A	p.G589R	Japanese	No	F	37	/	Cognitive impairment Gait disorder Speech disorder Apraxia Hemiplegia Tremor	Calcifications in the white matter Brain atrophy Abnormal signal in corpus callosum Dilation of the lateral ventricles	Konno T, 2016 (24)
13	c.1766G>A	p.G589E	USA	Yes	M	58	/	Cognitive impairment Psychiatric symptoms Urinary incontinence Seizures	Calcifications in the white matter Abnormal signal in frontal, parietal and	Konno T, 2016 (24)

								Parkinsonism Apraxia	temporal lobes Thinning of corpus callosum	
13	c.1766G>A	p.G589E	USA	Yes	F	47	58/11	Psychiatric symptoms Seizure Gait disorder	Bilateral white matter lesion Abnormal signal in frontal parietal lobes Brain atrophy CT: no calcifications	Fujioka S, 2013 (25)
13	c.1766G>A	p.G589E	USA	Yes	M	58	61/3	Cognitive impairment Psychiatric symptoms Seizures Gait disorder Tremor Urinary incontinence	Bilateral white matter lesion Abnormal signal in frontal parietal lobes Brain atrophy CT: no calcifications	Fujioka S, 2013 (25)
13	c.1786G>A	p. V596M	European	No	F	25	35/10	Cognitive impairment Psychiatric symptoms Seizures	Bilateral white matter lesion Abnormal signal in frontal parietal lobes	Lynch DS, 2016 (26)
13	c.1858+1G>T	/	Chinese	Yes	M	37	/	Speech disorder Cognitive impairment Tremor	Abnormal signal in lateral ventricle Abnormal signal in parietal lobe Bilateral white matter lesion	Yang X, 2019 (27)

14	/	p.L630R	European	No	M	45	52/7	Hemiplegia Parkinsonism	/	Guerreiro R, 2013 (20)
14	c.1901T>G	p.L634R	European	No	F	53	/	Hemiplegia Cognitive impairment Gait disorder	Abnormal signal in frontal, parietal and temporal lobes Thinning of corpus callosum Brain atrophy	Lynch DS, 2017 (28)
15	c.1954G>C	p.A652P	Japanese	No	M	30	/	Cognitive impairment Psychiatric symptoms Gait disorder	Abnormal signal in corpus callosum Bilateral white matter lesion Abnormal signal in frontal lobe Dilation of the lateral ventricles Calcifications in the white matter	Konno T, 2016 (24)
15	c.1957T>C	p.C653R	European	Yes	F	43	/	Cognitive impairment Hemiplegia, Dysphagia, Seizures Psychiatric symptoms	Brain atrophy Thinning of corpus callosum	Battisti C, 2014 (29)
15	c.1967G>A	p.C653 Y	Japanese	Yes	F	48	53/5	Hemiplegia, Seizures Cognitive impairment Apraxia	Abnormal signal in lateral ventricle Bilateral white matter	Riku Y, 2014 (30)

								Speech disorder	lesion Abnormal signal in frontal lobe Dilation of the lateral ventricles Brain atrophy	
15	c.1987G>A	p.E633K	European	No	/	29	/	Cognitive impairment Psychiatric symptoms Parkinsonism Apraxia	Bilateral white matter lesion Abnormal signal in frontal lobe Brain atrophy	Lynch DS, 2016 (26)
15	c.1987G>A	p.E633K	European	No	F	46	54/8	Cognitive impairment Psychiatric symptoms Seizures	Bilateral white matter lesion	Guerreiro R, 2013 (20)
15	c.1985T>C	p.I662T	USA	Yes	M	40	/	Cognitive impairment	Bilateral white matter lesion Abnormal signal in lateral ventricle CT: no calcifications	Miura T, 2018 (19)
15	c.1990G>A	p.E664K	American	Yes	F	35	/	Cognitive impairment Psychiatric symptoms Hemiplegia	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe Bilateral white matter lesion Dilation of the lateral	Eichler FS, 2016 (31)

									ventricles	
15	c.1990G>A	p.E664K	American	Yes	M	56	/	Cognitive impairment Hemiplegia Psychiatric symptoms Gait disorder Parkinsonism Seizures	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe Bilateral white matter lesion Dilation of the lateral ventricles	Eichler FS, 2016 (31)
15	c.1990G>A	p.E664K	American	Yes	F	54	60/6	Hemiplegia, Seizures Psychiatric symptoms Cognitive impairment Parkinsonism	Abnormal signal in lateral ventricle Bilateral white matter lesion Abnormal signal in frontal lobe Dilation of the lateral ventricles	Eichler FS, 2016 (31)
15	c.1990G>A	p.E664K	American	Yes	F	55	60/5	Hemiplegia, Seizures Psychiatric symptoms Cognitive impairment	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe Bilateral white matter lesion Abnormal signal in frontal lobe	Eichler FS, 2016 (31)

									Dilation of the lateral ventricles	
16	c.2060_2061insT	p.S688EfsX13	Japanese	No	F	41	54/13	Cognitive impairment Psychiatric symptoms	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe Abnormal signal in the corpus callosum Bilateral white matter lesion	Takuya K, 2014 (32)
17	/	p.S759F	Japanese	Yes	M	55	62/7	Cognitive impairment Psychiatric symptoms Parkinsonism	/	Kinoshita M, 2014 (33)
17	c.2287G>A	p. A763P	European	No	M	45	51/6	Psychiatric symptoms Parkinsonism Apraxia Seizures	Bilateral white matter lesion Abnormal signal in frontal, parietal and temporal lobes	Lynch DS, 2016 (26)
17	c.2294G>A	p.G765D	Japanese	Yes	F	37	/	Cognitive impairment Psychiatric symptoms Parkinsonism Gait disorder	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe Bilateral white matter lesion Abnormal signal in the corpus callosum	Takuya K, 2014 (32)

									Calcifications in the white matter	
17	c.2297T>	p.M766V	Korean	Yes	M	39	/	Cognitive impairment Speech disorder Tremor Psychiatric symptoms	Brain atrophy Thinning of corpus callosum	Kim EJ, 2015 (34)
17	c.2297T>C	p.M766T	European	Yes	F	18	/	Psychiatric symptoms Cognitive impairment Parkinsonism	Bilateral white matter lesion Abnormal signal in frontal lobe Brain atrophy Dilation of the lateral ventricles Thinning of the corpus callosum Calcifications in the white matter	Konno T, 2016 (24)
17	c. 2308G>C	p.A770P	European	Yes	M	52	63/11	Psychiatric symptoms Cognitive impairment Parkinsonism	/	Rademakers R, 2012 (22)
17	c.2320-2A>G	p.C774_N814del	USA	Yes	M	50	55/5	Psychiatric symptoms Cognitive impairment Parkinsonism Seizures	/	Rademakers R, 2012 (22)
18	c.2324T>A	p.I775N	USA	Yes	M	48	/	Psychiatric symptoms Gait disorder Parkinsonism	/	Rademakers R, 2012 (22)

18	c.2330G>A	p.R777Q	USA	Yes	F	40	/	Psychiatric symptoms Speech disorder Cognitive impairment	Abnormal signal in parietal lobe Abnormal signal in temporal lobe Abnormal signal in Brainstem Bilateral white matter lesion Abnormal signal in the corpus callosum CT: no calcifications	Makary MS, 2019 (35)
18	c.2330G>A	p.R777Q	European	Yes	F	22	23/1	Dysarthria, Dysphagia Cognitive impairment	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe Bilateral white matter lesion	Hoffmann S, 2014 (36)
18	c.2330G>A	p.R777Q	European	No	F	38	/	Cognitive impairment Parkinsonism Gait disorder Psychiatric symptoms	Brain atrophy Bilateral white matter lesion Abnormal signal in lateral ventricle Abnormal signal in frontal lobe	Karle KN, 2013 (37)
18	c.2330G>A	p.R777Q	European	Yes	F	60	65/5	Cognitive impairment Apraxia	Bilateral white matter lesion	Guerreiro R, 2013 (20)

								Speech disorder Seizures		
18	c.2330G>A	p.R777Q	European	Yes	M	44	48/4	Cognitive impairment Apraxia Speech disorder Parkinsonism Seizures	Bilateral white matter lesion	Guerreiro R, 2013 (20)
18	c.2330G>A	p.R777Q	European	Yes	M	58	/	Cognitive impairment	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe Brain atrophy	Guerreiro R, 2013 (20)
18	c.2330G>A	p.R777Q	Japanese	No	M	24	/	Dysphagia Hemiplegia Difficulty breathing	Bilateral white matter lesion Calcifications in the white matter Brain atrophy	Inui T, 2013 (38)
18	/	p.R777W	Japanese	Yes	M	53	56/3	Cognitive impairment Parkinsonism Psychiatric symptoms Apraxia Seizures	/	Kinoshita M, 2014 (33)
18	/	p.R777W	Japanese	Yes	M	40	/	Cognitive impairment Parkinsonism Psychiatric symptoms Apraxia	/	Kinoshita M, 2014 (33)

18	/	p.R777W	European	Yes	F	46	52/6	Cognitive impairment Seizures	/	Guerreiro R, 2013 (20)
18	c.2334C>A	p.D778E	USA	No	F	60	66/6	Cognitive impairment Hemiplegia	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe Bilateral white matter lesion Calcifications in the white matter	Miura T, 2018 (19)
18	c.2342C>A	p.A781E	Japanese	Yes	F	36	/	Cognitive impairment Psychiatric symptoms Parkinsonism	Abnormal signal in lateral ventricle Bilateral white matter lesion Abnormal signal in the corpus callosum Calcifications in the white matter	Takuya K, 2014 (32)
18	c.2342C>T	p.A781V	European	Yes	M	50	/	Apraxia Parkinsonism	Bilateral white matter lesion Abnormal signal in lateral ventricle Brain atrophy	Karle KN, 2013 (37)
18	c.2342C >T	p.A781V	European	Yes	M	48	/	Cognitive impairment Parkinsonism Gait disorder	Abnormal signal in lateral ventricle Bilateral white matter	Ahmed R, 2013 (39)

								Psychiatric symptoms	lesion Thinning of corpus callosum	
18	c.2342C >T	p.A781V	European	Yes	M	67	/	Hemiplegia Parkinsonism Cognitive impairment	Brain atrophy Dilation of the lateral ventricles Abnormal signal in parietal lobe Abnormal signal in lateral ventricle Bilateral white matter lesion Thinning of corpus callosum	Ahmed R, 2013 (39)
18	c.2342C >T	p.A781V	European	Yes	M	56	/	Psychiatric symptoms Gait disorder Cognitive impairment	Brain atrophy Abnormal signal in lateral ventricle Abnormal signal in frontal lobe Bilateral white matter lesion CT: no calcifications	Ahmed R, 2013 (39)
18	c.2342C >T	p.A781V	Korean	Yes	M	44	49/5	Cognitive impairment Hemiplegia Psychiatric symptoms Speech disorder	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe and parietal	Kim EJ, 2015 (34)

									lobe Bilateral white matter lesion Brain atrophy Thinning of corpus callosum	
18	c.2342C >T	p.A781V	Korean	Yes	F	41	/	Speech disorder Gait disorder Cognitive impairment Psychiatric symptoms	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe and parietal lobe Bilateral white matter lesion Brain atrophy Thinning of corpus callosum	Kim EJ, 2015 (34)
18	c.2345G>A	p.R782H	Korean	Yes	F	37	42/5	Speech disorder Dysarthria Gait disorder Cognitive impairment	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe and parietal lobe Bilateral white matter lesion Brain atrophy Thinning of corpus callosum	Kim EJ, 2015 (34)

18	c.2345G>A	p.R782H	Chinese	Yes	F	30	31/1	Hemiplegia Gait disorder Cognitive impairment Speech disorder	Abnormal signal in lateral ventricle Abnormal signal in parietal lobe Bilateral white matter lesion Thinning of corpus callosum	Shu Y, 2016 (40)
18	c.2345G>A	p.R782H	USA	Yes	F	Late 40s	56/6	Cognitive impairment Psychiatric symptoms Parkinsonism	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe Bilateral white matter lesion	Robinson JL, 2015 (41)
18	c.2345G>A	p.R782H	USA	Yes	F	54	57/3	Gait disorder Cognitive impairment Parkinsonism	Abnormal signal in lateral ventricle Bilateral white matter lesion	Robinson JL, 2015 (41)
18	c.2345G>A	p.R782H	USA	Yes	M	Early 50s	55/5	Gait disorder Cognitive impairment Parkinsonism	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe and parietal lobe Bilateral white matter lesion	Robinson JL, 2015 (41)
18	c.2345G>A	p.R782H	USA	Yes	F	51	/	Cognitive impairment	Abnormal signal in	Nicholson

								Psychiatric symptoms	temporal lobe Brain atrophy Bilateral white matter lesion CT: no calcifications	AM, 2013 (42)
18	c.2345G>A	p.R782H	Japanese	Yes	F	51	/	Cognitive impairment Seizures Psychiatric symptoms	Abnormal signal in temporal lobe Brain atrophy Bilateral white matter lesion	Kinoshita M, 2012 (43)
18	c.2345G>A	p.R782H	European	Yes	M	46	/	Cognitive impairment Psychiatric symptoms Apraxia	Abnormal signal in frontal, parietal and temporal lobes Thinning of corpus callosum Brain atrophy	Lynch DS, 2017 (28)
18	c.2344C>T	p.R782C	Chinese	Yes	M	45	/	Hemiplegia Psychiatric symptoms Parkinsonism Cognitive impairment	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe and parietal lobe Bilateral white matter lesion Dilation of the lateral ventricles	Yu X, 2017 (44)
18	/	p.R782G	European	Yes	M	40	41/1	Hemiplegia	/	Foulds N,

								Cognitive impairment Psychiatric symptoms Speech disorder Seizures		2015 (45)
18	/	p.R782G	European	Yes	F	38	40/2	Hemiplegia Cognitive impairment Psychiatric symptoms Urinary incontinence	Bilateral white matter lesion	Foulds N, 2015 (45)
18	/	p.R782G	European	Yes	M	57	59/2	Psychiatric symptoms Gait disorder Urinary incontinence Seizures	Thinning of corpus callosum Brain atrophy Abnormal signal in frontal, parietal lobes	Foulds N, 2015 (45)
18	c.2350G>A	p.V784M	Canadian	Yes	F	39	/	Psychiatric symptoms Cognitive impairment Gait disorder	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe Bilateral white matter lesion Dilation of the lateral ventricles	Roberta LP, 2014 (46)
18	c.2375C>A	p.A792D	Japanese	Yes	M	41	/	Cognitive impairment Psychiatric symptoms Hemiplegia	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe and parietal lobe	Ueda S, 2015 (47)

									Bilateral white matter lesion Calcifications in the white matter	
18	c.2378A>C	p.K793T	Japanese	No	M	40	41/1	Cognitive impairment Gait disorder Hemiplegia Dysarthria	Abnormal signal in lateral ventricle Abnormal signal in parietal lobe Bilateral white matter lesion Brain atrophy Dilation of the lateral ventricles	Yasufumi K, 2013 (48)
18	c.2381T>C	p.I794T	European	No	F	29	/	Gait disorder Cognitive impairment Dysarthria	Abnormal signal in lateral ventricle Bilateral white matter lesion Abnormal signal in the corpus callosum, calcifications in white matter	Meyer-Ohlendorf M, 2015 (49)
18	c.2381T>C	p.I794T	Chinese	Yes	F	32	/	Speech disorder Tremor, Gait disorder Psychiatric symptoms Cognitive impairment	Abnormal signal in lateral ventricle Bilateral white matter lesion calcifications in white	Lan MY, 2015 (50)

									matter	
18	c.2381T>C	p.I794T	Japanese	Yes	F	20	/	Hemiplegia	Abnormal signal in lateral ventricle Abnormal signal in parietal lobe Bilateral white matter lesion	KitaniMorii F, 2013 (51)
18	c.2381T>C	p.I794T	Japanese	No	M	40	/	Cognitive impairment Psychiatric symptoms Seizures Parkinsonism	Abnormal signal in lateral ventricle Bilateral white matter lesion Abnormal signal in the corpus callosum Calcifications in the white matter	Takuya K, 2014 (32)
18	c.2381T>C	p.I794T	Japanese	Yes	F	55	/	Cognitive impairment Psychiatric symptoms Parkinsonism Seizures	Abnormal signal in lateral ventricle Bilateral white matter lesion Abnormal signal in the corpus callosum Calcifications in the white matter	Takuya K, 2014 (32)

18	c.2381T>C	p.I794T	European	Yes	F	40	43/3	Psychiatric symptoms Hemiplegia Seizures Gait disorder	Bilateral white matter lesion Thinning of corpus callosum	Karle KN, 2013 (37)
18	c.2381T>C	p.I794T	Chinese	Yes	F	42	/	Psychiatric symptoms Parkinsonism Urinary incontinence Cognitive impairment	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe and parietal lobe Bilateral white matter lesion	Wu L, 2017 (18)
18	c.2381T>C	p.I794T	Japanese	No	F	28	/	Gait disorder Cognitive impairment Dysarthria Dysphagia Urinary incontinence	Abnormal signal in parietal lobe Abnormal signal in lateral ventricle Bilateral white matter lesion Thinning of corpus callosum	Saitoh BY, 2013 (52)
18	c.2381T>C	p.I794T	Chinese	Yes	F	42	/	Cognitive impairment Psychiatric symptoms Parkinsonism Urinary incontinence	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe Bilateral white matter lesion Thinning of corpus	Bai Y, 2018 (53)

									callosum	
18	c.2381T>C	p.I794T	Japanese	No	F	27	/	Cognitive impairment Psychiatric symptoms Seizures Apraxia	/	Kinoshita M, 2014 (33)
18	c.2381T>C	p.I794T	European	No	M	52	/	Cognitive impairment Parkinsonism Hemiplegia	Abnormal signal in frontal, parietal and temporal lobes Thinning of corpus callosum Brain atrophy	Lynch DS, 2017 (28)
18	c.2381T >C	p.I794T	European	No	M	46	49/3	Cognitive impairment	Bilateral white matter lesion	Guerreiro R, 2013 (20)
18	c.2380A>T	p.I794F	USA	No	M	56	/	Cognitive impairment Parkinsonism	Bilateral white matter lesion Abnormal signal in lateral ventricle Abnormal signal in frontal lobe and parietal lobe Calcifications in the white matter	Miura T, 2018 (19)
18	c.2442+1G>T	/	Japanese	No	M	53	57/4	Cognitive impairment Psychiatric symptoms Parkinsonism	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe	Takuya K, 2014 (32)

									Bilateral white matter lesion Abnormal signal in the corpus callosum Calcifications in the white matter	
18	c.2442+1G>T	/	Korean	No	F	47	/	Cognitive impairment Speech disorder Parkinsonism Apraxia Seizures	Bilateral white matter lesion Thinning of the corpus callosum white matter lesions Brain atrophy Abnormal signal in lateral ventricle Abnormal signal in frontal lobe and parietal lobe CT: no calcifications	Lee D, 2015 (54)
18	c.2442+1G>A	/	European	No	/	43	/	Psychiatric symptoms Parkinsonism Apraxia	Abnormal signal in frontal, parietal and temporal lobes thinning of the corpus callosum white matter lesions Brain atrophy	Lynch DS, 2016 (26)
18	c.2442+2T>C	/	Japanese	Yes	M	44	53/9	Gait disorder, tremor, Cognitive impairment	Abnormal signal in front lobe and lateral ventricle	Kawakami I, 2016 (55)

								Psychiatric symptoms Parkinsonism Dysarthria Dysphagia	Bilateral white matter lesion Thinning of corpus callosum Brain atrophy	
18	c.2442+2T>C	/	Japanese	Yes	M	38	/	Hemiplegia Cognitive impairment Speech disorder Dysarthria, dysphagia Parkinsonism Seizures Urinary incontinence Gait disorder	Bilateral white matter lesion Thinning of corpus callosum Brain atrophy	Kawakami I, 2016 (55)
18	c.2442+5G>A	/	Japanese	Yes	F	27	/	Hemiplegia Cognitive impairment Gait disorder Urinary incontinence Apraxia	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe and parietal lobe Bilateral white matter lesion Abnormal signal in corpus callosum Calcifications in the white matter	Konno T, 2016 (24)
18	c.2442+5G>A	/	Japanese	Yes	M	58	/	Cognitive impairment Psychiatric symptoms	Abnormal signal in lateral ventricle	Konno T, 2016 (24)

									Abnormal signal in frontal lobe Bilateral white matter lesion Abnormal signal in corpus callosum Calcifications in the white matter	
18	c.2442+5G>C	/	USA	No	F	23	/	Cognitive impairment Psychiatric symptoms Gait disorder Apraxia	Calcifications in the white matter	Konno T, 2016 (24)
18	c.2350T>C	p.L817P	USA	No	M	21	36/15	Cognitive impairment Headache	/	Guerreiro R, 2013 (20)
19	c.2467C>T	p.A823V	Japanese	Yes	F	51	/	Cognitive impairment	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe and parietal lobe Bilateral white matter lesion Brain atrophy Calcifications in the white matter	Terasawa Y, 2013 (56)
19	c.2470C>T	p.P824S	Japanese	No	F	45	/	Cognitive impairment Psychiatric symptoms	Abnormal signal in lateral ventricle	Takuya K, 2014 (32)

								Parkinsonism	Abnormal signal in frontal lobe lobe Bilateral white matter lesion Abnormal signal in the corpus callosum Calcifications in the white matter	
19	c.2473G>A	p. E825K	European	Yes	F	42	/	Cognitive impairment Apraxia Tremor	Thinning of the corpus callosum Bilateral white matter lesion Abnormal signal in front lobe Brain atrophy	Lynch DS, 2016 (26)
19	/	p.I827T	European	Yes	F	42	/	Tremor Gait disorder	Bilateral white matter lesion	Guerreiro R, 2013 (20)
19	c.2775T>C	p.F828S	USA	No	F	39	/	Cognitive impairment Psychiatric symptoms Hemiplegia	Bilateral white matter lesion Thinning of corpus callosum	Kleinfeld K, 2013 (57)
19	c.2509G>T	p.D837Y	USA	Yes	F	43	50/7	Cognitive impairment Gait disorder	Bilateral white matter lesion Brain atrophy	Rademakers R, 2012 (22)
19	c.2512G>C	p.V838L	European	No	M	53	/	Cognitive impairment Parkinsonism	Bilateral white matter lesion	Karle KN, 2013 (37)

								Seizures Gait disorder	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe and parietal lobe Brain atrophy	
19	c.2527_2530delinsGGCA	p.I843_L844delinsGI	German	Yes	F	43	/	Gait disorder, Hemiplegia, Apraxia, parkinsonism, Nystagmus Speech disorder	Abnormal signal in lateral ventricle Abnormal signal in parietal lobe Bilateral white matter lesion Thinning of corpus callosum Brain atrophy	Kraya T, 2019 (58)
19	c.2527A>T	p.I843F	European	Yes	M	55	/	Psychiatric symptoms Seizures	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe and parietal lobe Bilateral white matter lesion Brain atrophy CT: no calcifications	Battisti C, 2014 (29)
19	c.2528T>A	p.I843N	European	No	F	40	/	Cognitive impairment Parkinsonism	Bilateral white matter lesion	Karle KN, 2013 (37)

								Gait disorder Psychiatric symptoms	Brain atrophy Thinning of corpus callosum	
19	c.2539G>A	p.E847K	European	No	M	35	/	Psychiatric symptoms Cognitive impairment Hemiplegia	Abnormal signal in lateral ventricle Abnormal signal in frontal lobe and parietal lobe Bilateral white matter lesion Dilation of the lateral ventricles Thinning of corpus callosum	Ilaria DD, 2015 (59)
19	/	p.E847D	European	Yes	F	44	/	Cognitive impairment Hemiplegia	/	Guerreiro R, 2013 (20)
19	c.2540A>T	p.E847V	USA	No	F	33	/	Cognitive impairment Apraxia Urinary incontinence Nystagmus, Speech disorder	Bilateral white matter lesion Thinning of corpus callosum Abnormal signal in lateral ventricle and front lobe CT: no calcifications	Gore E, 2016 (60)
19	c.2546T>C	p.F849S	Chinese	No	F	58	/	Cognitive impairment Psychiatric symptoms Urinary incontinence	Abnormal signal in lateral ventricle and frontal lobe	Lan MY, 2015 (50)

								Parkinsonism	Bilateral white matter lesion Thinning of corpus callosum CT: no calcifications	
19	c.2546_2548delTCT	p.F849del	USA	Yes	F	63	67/4	Cognitive impairment Parkinsonism Gait disorder Seizures	/	Rademakers R, 2012 (22)
20	c.2562T>A	p.N854K	European	Yes	F	44	49/5	Cognitive impairment Nystagmus, dysphagia Parkinsonism Gait disorder Urinary incontinence	Abnormal signal in lateral ventricle Bilateral white matter lesion Thinning of corpus callosum	Granberg T, 2016 (61)
20	c.2562T>A	p.N854K	European	Yes	M	47	51/4	Cognitive impairment Psychiatric symptoms Hemiplegia Gait disorder	Abnormal signal in lateral ventricle and frontal lobe Bilateral white matter lesion Thinning of corpus callosum	Granberg T, 2016 (61)
20	c.2562T>A	p.N854K	European	Yes	M	50	53/3	Cognitive impairment Psychiatric symptoms Hemiplegia Gait disorder	Abnormal signal in lateral ventricle and frontal lobe Bilateral white matter lesion Thinning of corpus	Granberg T, 2016 (61)

									callosum	
20	c.2562T>A	p.N854K	European	Yes	F	54	69/15	Cognitive impairment Parkinsonism Gait disorder Dysphagia Nystagmus	Abnormal signal in lateral ventricle and frontal lobe Bilateral white matter lesion Thinning of corpus callosum	Granberg T, 2016 (61)
20	c.2562T>A	p.N854K	European	Yes	M	56	61/5	Cognitive impairment Parkinsonism Gait disorder Dysphagia Nystagmus	Abnormal signal in lateral ventricle and frontal lobe Bilateral white matter lesion Thinning of corpus callosum	Granberg T, 2016 (61)
20	c.2562T>A	p.N854K	European	Yes	F	29	/	Hemiplegia	Abnormal signal in lateral ventricle Abnormal signal in lateral ventricle and frontal lobe Bilateral white matter lesion	Sundal C, 2015 (62)
20	c.2563C>A	p.P855T	Chinese	Yes	F	41	/	Cognitive impairment Gait disorder Urinary incontinence	Bilateral white matter lesion Thinning of corpus callosum Dilation of the lateral	Cheng X, 2015 (63)

									ventricles Brain atrophy	
20	c.2563C>A	p.P855T	Chinese	Yes	M	34	/	Cognitive impairment	/	Cheng X, 2015 (63)
20	c.2563C>A	p.P855T	Chinese	Yes	F	50	51/1	Hemiplegia Gait disorder Speech disorder	/	Cheng X, 2015 (63)
20	c.2563C>A	p.P855T	Chinese	Yes	F	30	/	Psychiatric symptoms	/	Cheng X, 2015 (63)
20	/	p.Y856H	European	Yes	F	47	/	Cognitive impairment Gait disorder Dysarthria Speech disorder	Bilateral white matter lesion Abnormal signal in frontal, parietal lobe	Guerreiro R, 2013 (20)
20	/	p.Y856H	European	Yes	F	39	42/3	Cognitive impairment Gait disorder Dysarthria Seizures	Bilateral white matter lesion	Guerreiro R, 2013 (20)
20	/	p.Y856H	European	Yes	F	42	/	Cognitive impairment Apraxia Parkinsonian seizures	/	Guerreiro R, 2013 (20)
20	/	p.Y856H	European	Yes	F	42	/	Cognitive impairment	/	Guerreiro R, 2013 (20)
20	/	p.Y856H	European	Yes	F	55	64/9	Parkinsonism Dysarthria Cognitive impairment	/	Guerreiro R, 2013 (20)
20	c.2570C>T	p.P857L	European	No	F	38	/	Gait disorder	Bilateral white matter	Lynch DS,

								Hemiplegia Cognitive impairment	lesion Abnormal signal in frontal, parietal and temporal lobes Thinning of corpus callosum Brain atrophy	2017 (28)
20	c.2603T>C	p.L868P	USA	Yes	F	55	63/8	Cognitive impairment Psychiatric symptoms Parkinsonism Seizures	/	Rademakers R, 2012 (22)
20	c.2624T>C	p.M875T	USA	Yes	M	58	66/8	Cognitive impairment Psychiatric symptoms Parkinsonism Seizures	/	Rademakers R, 2012 (22)
20	c.2624T>C	p.M875T	USA	Yes	M	71	/	Cognitive impairment Psychiatric symptoms	/	Rademakers R, 2012 (22)
20	c.2624T>C	p.M875T	USA	Yes	M	41	43/2	Cognitive impairment Psychiatric symptoms Parkinsonism	/	Rademakers R, 2012 (22)
20	c.2624T>C	p.M875T	USA	Yes	M	46	49/3	Cognitive impairment Psychiatric symptoms Parkinsonism Speech disorder	/	Rademakers R, 2012 (22)
21	c.2629C>T	p.Q877X	European	Yes	M	28	/	Cognitive impairment Psychiatric symptoms Parkinsonism	Bilateral white matter lesion Thinning of corpus	Karle KN, 2013 (20)

								Gait disorder	callosum	
21	c.2655-2A>G	/	European	Yes	F	50	52/2	Hemiplegia Cognitive impairment	/	Guerreiro R, 2013 (20)
21	c.2632C>T	p.P878S	USA	No	M	45	/	Cognitive impairment Parkinsonism Seizures	Bilateral white matter lesion Abnormal signal in lateral ventricle Calcifications in the white matter	Miura T, 2018 (19)
21	c.2632C>G	p.P878A	USA	Yes	M	57	64/7	Cognitive impairment Parkinsonism Psychiatric symptoms	Bilateral white matter lesion Thinning of corpus callosum CT no calcifications	Miura T, 2018 (19)
21	c.2632C>A	p.P878T	USA	Yes	F	39	49/10	Cognitive impairment Seizures Psychiatric symptoms	/	Rademakers R, 2012 (22)
21	c.2632C>A	p.P878T	USA	Yes	M	33	43/10	Cognitive impairment Seizures Psychiatric symptoms	/	Rademakers R, 2012 (22)
21	c.2655-2A>G	/	European	Yes	F	50	52/2	Cognitive impairment Seizures Speech disorder	/	Guerreiro R, 2013 (20)
21	c.2655_2656delAT	p.T886Qfs*55	USA	No	M	46	/	Cognitive impairment	Calcifications in the white matter	Miura T, 2018 (19)
21	c.2699G>A	p.R900K	European	Yes	M	53	/	Hemiplegia	Abnormal signal in	Körtvelyessy

								Cognitive impairment	lateral ventricle Abnormal signal in parietal lobe Bilateral white matter lesion Abnormal signal in spinal cord	P, 2015 (64)
21	/	p.P901S	USA	No	/	20	45/25	Cognitive impairment	/	Guerreiro R, 2013 (20)
21	c.2717T>C	p.I906T	European	NA	M	36	/	Hemiplegia Cognitive impairment Urinary incontinence Dysphagia	Abnormal signal in lateral ventricle and frontal lobe Bilateral white matter lesion	Battisti C, 2014 (29)
22	c.2909_2910ins ATCA	p.970Sfs*108	Chinese	No	M	30	/	Cognitive impairment Urinary incontinence Psychiatric symptoms Hemiplegia	Abnormal signal in lateral ventricle Bilateral white matter lesion Abnormal signal in frontal lobe Abnormal signal in the corpus callosum	Wu L, 2017 (18)

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