Supplementary Table 1a. HD-like syndromes with typical onset in adulthood.

	Demographic features	**Core neurological features**	**Cognitive and behavioural abnormalities**	**Ancillary investigations**	**Molecular genetics**
HDL-1	Prevalence: unknown; Observed only in Caucasian families	Chorea, rigidity, ataxia, dysarthria, seizures	Cognitive deterioration, personality changes, mania-like symptoms	MRI: atrophy of frontal and temporal lobes, cerebellum and basal ganglia	AD *PRNP* gene: 192-nucleotide insertion leading to extra-octapeptide repeats in the prion protein
HDL-2	Prevalence unknown; South African ancestry	Chorea or parkinsonism (2 forms), dysarthria, hyperreflexia, jerky	Dementia, depression, apathy, irritability	MRI: atrophy of caudate and cortex Acanthocytosis	AD *JPH3* gene (TMA): CTG repeat

		saccades			expansion
SCA17 (HDL-4)	Prevalence 0.5-1.6/million All ethnic groups	Ataxia, chorea, dystonia, eye movement abnormalities, pyramidal signs, rigidity	Dementia, depression, apathy, psychosis	MRI: atrophy of cerebellum, brainstem, caudate, cortex	AD *TBP* gene (TMA): CAA/CAG repeat expansion
DRPLA	Prevalence 4.8/million in the Japanese population; much rarer in other ethnic groups	Ataxia, chorea, dystonia, myoclonus, epilepsy	Dementia or progressive intellectual deterioration (in children), personality changes, psychosis	MRI: atrophy of cerebellum and brainstem (esp. pontine tegmentum)	AD *ATN1* gene (TMA): CAG repeat expansion
Chorea-Acanthocytosis	Prevalence unknown All ethnic groups	Chorea, dystonia, dysarthria, dysphagia, tongue/lip biting, seizures,		MRI: atrophy of caudate and T_2-weighted signal increase in caudate and putamen	AR *VPS13A*: Sequence analysis or deletion-

		myopathy, sensorimotor axonopathy		Acanthocytosis Raised serum CK	duplication analysis
McLeod syndrome	Prevalence unknown All ethnic groups	Chorea, seizures, sensorimotor axonopathy, myopaty, dilated CMP and arrhythmias	Mild cognitive deficits, personality disorder, anxiety, depression, OCD, bipolar or schizo-affective disorder	MRI: atrophy of caudate and putamen McLeod blood group phenotype (Kx erythrocyte antigen) Acanthocytosis Compensated hemolysis Raised serum CK, LDH, AST, ALT Abnormal EMG/NCS	XR *XK*: Direct DNA methods (available only for research)

				Muscle CT or biopsy: fatty degeneration of lower leg muscles Cardiac abnormalities on echo and ECG	
Neuro-ferritinopathy	Prevalence unknown Possible common founder in Europe	Dystonia, chorea, parkinsonism, dysarthria, dysphonia, dysphagia, hyperreflexia	Mild cognitive difficulties with cognitive and behavioural changes only later on in the disease course	MRI: iron deposition in basal ganglia, cystic degeneration Low serum ferritin levels	AD *FTL*: Sequence analysis detecting point mutations and small deletions or insertions in 80% of familial cases (much less in

					sporadic presentations)
SCA14	Prevalence unknown Different ethnic groups	Ataxia, dysarthria, dysphagia, dysphonia, eye movement abnormalities , sensory loss, pyramidal signs, chorea	Dementia, depression	MRI: Mild-to-moderate cerebellar atrophy	AD *PRKCG*: Sequence analysis detecting point mutations and small deletions (unknown detection rate)
SCA8	Prevalence unknown May be especially common in Finland	Ataxia, dysarthria, hyperreflexia, chorea	Cognitive deterioration	MRI: mild-to-moderate cerebellar atrophy	AD *SCA8/ATXN 8*: Trinucleotide repeat expansion within two

overlapping genes

AD: autosomal dominant; TMA: targeted mutation analysis; AR: autosomal recessive; XR: X-linked recessive; CMP: cardiomyopathy.

Supplementary Table 1b. HD-like syndromes with typical onset in the first two decades.

	Demographic features	**Core neurological features**	**Cognitive and behavioural abnormalities**	**Ancillary investigations**	**Molecular genetics**
Benign hereditary chorea	Prevalence: unknown; Observed all ethnic groups	Chorea, very rarely dysarthria, dystonia, myoclonus	Psychosis very rare	None	AD *NKX2-1* gene in a proportion of patients: point mutations or large deletion
Friedreich ataxia	Prevalence 20-40/million Well documented in Europe, Middle East, India, North Africa	Ataxia, areflexia, dysarthria, loss in position and/or vibration sense, pyramidal signs, chorea, scoliosis, pes	Mild abnormalities of executive functioning	MRI: atrophy of cervical spinal cord and cerebellum later on in the course Glucose tolerance test	AR *FXN* gene (TMA): GAA repeat expansion

	(not in Southeast Asia, sub-saharan Africa, Native Americans)	cavus, cardiomyopathy, optic atrophy, deafness, glucose intolerance		abnormal Echocardiography and ECG abnormalities Abnormal NCS and central motor conduction time	Sequence analysis or deletion- duplication analysis also possible
Ataxia-teleangiectasia	Prevalence 10-20/million live births All ethnic groups	Ataxia, head tilting, dysarthria, teleangiectasias, oculomotor apraxia, chorea, dystonia, immunodeficiency and increased rate of infections (especially respiratory tract) and neoplasms	Learning disabilities possible	Raised serum AFP Severe depletion of ATM protein on immunoblotting or other cell-based functional assays	AR *ATM* gene (TMA): c.103C>T (common allele in specific ethnic populations) Sequence, deletion-duplication

					and haplotype analyses also possible
Ataxia with oculomotor apraxia types 1 and 2	Type 1: Prevalence 0.041/million in Portugal (unknown elsewhere) All ethnic groups Type 2: approx. 2/million in Alsace (unknown elsewhere);	Ataxia, oculomotor apraxia, axonal neuropathy, chorea, dystonia	Cognitive impairment (different degrees)	MRI: atrophy of cerebellum Abnormal NCS Hypercholesterolemia Hypoalbuminemia (type 1) Raised serum AFP and CK (type 2) Elevated IgG and IgA levels (type 2)	Type 1: AR *APTX* gene (type 1) *SETX* gene (type 2): Sequence or deletion-duplication analyses

	mainly French-Canadian and Anglo-Norman populations				
PKAN	Prevalence estimated to approximately 1-3/million All ethnic groups	Dystonia, rigidity, dysarthria, dysphagia, eye movement abnormalities, chorea, gait disorder, pyramidal signs, retinal degeneration or optic atrophy		MRI: 'eye-of-the-tiger' sign in globus pallidus Acanthocytosis Low or absent plasma pre-beta lipoprotein fraction	AR *PANK2* gene: Sequence analysis
***PLA2G6*-associated neurodegeneration**	Prevalence unknown Different ethnic groups	Dystonia, parkinsonism (L-dopa responsive), pyramidal signs, dysphagia, dysarthria, optic atrophy,	Dementia or global developmental delay	MRI: iron deposition in globus pallidus or normal	AR *PLA2G6* gene: Direct DNA methods (available only for

		cerebellar features, chorea, sensorimotor axonopathy			research)
Kufor Rakeb syndrome	Prevalence unknown Different ethnic groups	Parkinsonism (L-dopa responsive), dysarthria, lip/chin myoclonic/tremor, dysphonia, dysphagia, pyramidal signs, supranuclear gaze palsy	Dementia, hallucinations, aggressive behaviour	MRI: iron deposition in basal ganglia or normal	AR *ATP13A2* gene: Direct DNA methods (available only for research)
Wilson's disease	Prevalence 30-35/million All ethnic groups, higher in China, Japan, and Sardinia	Tremor, loss of fine motor control, chorea, dystonia, rigidity, Kaiser-Fleischer rings, liver disease	Depression, anxiety, compulsions, phobias, personality changes, cognitive impairment	MRI: copper accumulation in putamen and globus pallidus Low serum ceruloplasmin, increased basal urinary copper	AR *ATP7B* gene (TMA): Sequence analysis and deletion-duplication

				excretion, increased hepatic copper concentration	testing also available
Aceruloplasminemia	Prevalence estimated as approximately 0.5/million in Japan Also other ethnic groups	Ataxia, dysarthria, nystagmus, dystonia, tremor, chorea, parkinsonism, diabetes mellitus	Cognitive deterioration	MRI: iron accumulation in basal ganglia Serum ceruloplasmin undetectable on Western blot, low serum copper, iron and ferritin, plasma ceruloplasmin ferroxidase activity undetectable Raised iron levels in liver and pancreatic	AR *CP* gene: Sequence analysis (available only for research)

islet beta cells

AD: autosomal dominant; TMA: targeted mutation analysis; AR: autosomal recessive.