Disease	**Gene** (loci)	**Normal CAG range**	**Disease CAG range** (full penetrance)	**Somatic expansion observed?**	**Major Neuropathology** (cell-types specified where known)	**Clinical Phenotype** (varies according to size of CAG repeat)
Huntington's Disease (HD)	HTT (autosomal)	6-35	40-250	Yes	Striatal and cortical atrophy with degeneration of striatal medium spiny neurons and cortical projection neurons	Progressive motor disability including chorea; cognitive decline, psychiatric features
Spinocerebellar ataxia type 1 (SCA1)	*ATXN1 (autosomal)*	6-35	>39 (no CAT) 45-81 (with CAT)	Yes	Brainstem atrophy; cerebellar atrophy and gliosis with Purkinje cell degeneration	Progressive cerebellar ataxia, spasticity, ophthalmoplegia, sensory and bulbar symptoms
Spinocerebellar ataxia type 2 (SCA2)	*ATXN2 (autosomal)*	14-31	37-270	Yes	Brainstem atrophy; cerebellar atrophy and degeneration of Purkinje and granule cells; neuronal degeneration and gliosis of the inferior olive and pons	Progressive cerebellar ataxia, slow eye saccades, peripheral neuropathy, resting tremor, myoclonus, dementia, sensory symptoms, occasional pure parkinsonism
Spinocerebellar ataxia type 3 (SCA3) / Machado-Joseph disease (MJD)	*ATXN3 (autosomal)*	12-44	~60-87	Yes	Pontine and spinal atrophy; cerebellar atrophy; degeneration of the spinocerebellar tract	Progressive cerebellar ataxia, spasticity, diplopia, ophthalmoplegia, bulging eyes, nystagmus, pyramidal signs, dystonic-rigid extrapyramidal syndrome, peripheral neuropathy, facial myokymia, sensory symptoms
Spinocerebellar ataxia type 6 (SCA6)	*CACNA1A (autosomal)*	≤18	20-33	No	Cerebellar atrophy with degeneration of Purkinje and granule cells	Progressive cerebellar ataxia, nystagmus

Spinocerebellar ataxia type 7 (SCA7)	*ATXN7 (autosomal)*	7-27	37-460	Yes	Cerebellar and brainstem atrophy; degeneration of retinal photoreceptors; neuronal loss and reactive gliosis in the cerebellar cortex, dentate nucleus, inferior olive, and pontine nuclei	Progressive cerebellar ataxia, impaired vision, ophthalmoplegia, spasticity, pyramidal signs
Spinocerebellar ataxia type 17 (SCA17)	*TBP (autosomal)*	25-40	49-66	Yes	Atrophy of the cortex, striatum, and cerebellum, with neuronal loss in the striatum and cerebellar Purkinje cell layer	Progressive cerebellar ataxia, spasticity, dementia, dystonia, chorea, parkinsonism, psychiatric symptoms, pyramidal signs
Spinal and Bulbar Muscular Atrophy (SBMA) / Kennedy's Disease	*AR* (X-linked)	<34	38-70	Yes	Degeneration of lower motor neurons in the spinal cord and bulbar region of brain stem	Neurogenic muscle atrophy and weakness, fasciculations, gynaecomastia, testicular atrophy, reduced fertility
Dentatorubral-pallidoluysian atrophy (DRPLA)	*ATN1 (autosomal)*	6-35	48-93	Yes	Degeneration of the dentatorubral and pallidoluysian systems; cerebellar and brainstem atrophy and cerebral white matter lesions	Ataxia, progressive myoclonus epilepsy, dystonia, chorea, intellectual deterioration (children), dementia (adults)