

Title: *VPS13A* Disease *GeneReview* Neuropathology

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There is some evidence of macroscopic bilateral atrophy of the caudate nucleus, the putamen, and the globus pallidus, corresponding to histologic loss of neurons and gliosis, which is particularly severe in the caudate but less so in the putamen and the external and internal pallidum [Connolly et al 2014, Liu et al 2018]. There is also a distinct cortical neurodegeneration [Liu et al 2019]. Pronounced neuronal loss in the substantia nigra is the likely neuropathologic correlate of parkinsonism. Gliosis and extraneuronal pigment, but no Lewy bodies, are observed in the substantia nigra. Hippocampal sclerosis likely correlates with temporal lobe seizures [Mente et al 2017]. The locus coeruleus, inferior olives, and cerebellum appear unaffected. Loss of spinal cord anterior horn cells, a correlate of neurogenic muscle atrophy, is seen in some of the autopsies of individuals with *VPS13A* disease. Gliosis may also occur in the thalamus.

References

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