**On-line Table: Summary of major inherited diseases with dentate nucleus involvement**

| Disease | Incidence | Genetic Mutation | Inheritance Pattern | Classic Clinical Syndrome | Dentate Appearance (CT/MRI) | Other Areas Affected |
|---|---|---|---|---|---|---|
| NBIA | 1/1,000,000 | 10 types | 8 AR<br>1 AD<br>1 X-linked dominant | Extrapyramidal abnormalities and cognitive dysfunction | CT: NA<br>MRI: iron deposition visualized as hypointensity on SWI/T2*GRE and T2WI | Iron deposition in the basal ganglia, thalami, substantia nigra |
| Fahr disease | 1/1,000,000 | *SLC20A2, PDGFRB, PDGFB, XPR1* | AD or AR | Progressive extrapyramidal symptoms and cognitive impairment | CT: dense symmetric calcification<br>MRI: mineralization will be hypointense on SWI/T2*GRE and T2WI; variable to hyperintense on T1WI depending on stage of disease and calcium metabolism | Dense calcification in the basal ganglia, thalamus, subcortical white matter |
| Leigh disease | 1/50,000 | *SURF1* | AR or X-linked | Developmental delay and regression | CT: Low attenuation<br>MRI: hyperintensity on T2WI with diffusion restriction<br>MRS: elevated choline and lactate levels | T2 hyperintensity and restricted diffusion of basal ganglia, thalamus, periaqueductal gray, substantia nigra, pons, medulla |
| Friedreich ataxia | 2/100,000 | *FXN* | AR | Progressive gait ataxia | CT/MRI: no visible abnormalities | Wallerian degeneration of the SCP; thinning of the cervical spinal cord |
| Canavan disease | 1/100,000 | *ASPA* | AR | Developmental delay, poor muscle tone | CT: low attenuation<br>MRI: T2 hyperintensity and diffusion restriction<br>MRS: elevated NAA | Diffuse atrophy and white matter changes; sparing of the putamen; variable involvement of thalamus, brain stem, and pons |
| Glutaric aciduria type 1 | 1/100,000 | *GCDH* | AR | Rapid neurologic deterioration following febrile illness | CT: low attenuation<br>MRI: T2 hyperintensity and diffusion restriction | Cystlike widening of the Sylvian fissures; frontotemporal atrophy<br>Subdural hematomas and hygromas |
| Maple syrup urine disease | 1/185,000 | *BCKDHA, BCKDHB, DBT* | AR | Failure to thrive and neurologic decline in the first few days of life | CT: Low attenuation<br>MRI: T2 hyperintensity and diffusion restriction<br>MRS: leucine, isoleucine, and valine | Generalized cerebral edema with marked involvement of the 4 DCN, brain stem, cerebral peduncles, and internal capsule |

**Note:**—NBIA indicates neurodegeneration with brain iron accumulation; NA, not applicable; DCN, deep cerebellar nuclei; AR, autosomal recessive; AD, autosomal dominant; GRE, gradient recalled-echo; SCP, superior cerebellar peduncle.