Supplementary Material

A Practical Approach to Early-Onset Parkinsonism

Supplementary Table 1. Imaging studies in different forms of EO parkinsonism.
Findings in IofIoflupane I 123 SPECT, Iodobenzamide I 123 SPECT, and Fluorodeoxyglucose F18 PET for the conditions are listed in the table, where available. NA, not available.

Disorder	*Ioflupane I 123 SPECT*	*Iodobenzamide I 123 SPECT*	*Fluorodeoxyglucose F18 PET*
PARK-PRKN	Abnormal[1]	NA	Normal[2]
PARK-PINK1	Abnormal[1]	Normal[3]	Probably normal[4]
PARK-FBXO7	Abnormal[5]	Normal[5]	Cortical hypometabolism[6]
PARK-RAB39B	Abnormal[7]	Abnormal[7]	NA
PARK-PTRHD1	Abnormal[8]	NA	NA
PARK-SYNJ1	Abnormal[9]	NA	Cortical and caudate hypometabolism[9,10]
PARK-DNAJC6	Abnormal[11]	NA	Normal[12]
PARK-VPS13C	Abnormal[13]	NA	Cortical hypometabolism[3]
PARK-SNCA	Abnormal[1]	NA	Cortical hypometabolism[14]
PARK-LRRK2	Abnormal[1]	NA	Mild parietal hypometabolism[15]
PARK-VPS35	Abnormal[16]	NA	NA
PARK-GBA	Abnormal[1]	NA	Normal or reduced uptake in basal ganglia, cortical parietoccipital hypometabolism[17,18]
Wilson disease	Abnormal[19]	Abnormal[20]	Cortical and striatal hypometabolism[21]
PKAN	Probably normal[22]	Probably normal[23]	NA
PLA2G6	Abnormal[24]	NA	Cortical hypometabolism[25]
MPAN	NA	NA	Cortical hypometabolism [26]
BPAN	Abnormal [27]	NA	Cortical hypometabolism [28]
Kufor-Rakeb Syndrome	Abnormal [29]	NA	NA
Fahr disease	Abnormal in some cases [30]	NA	Reduced uptake in basal ganglia in some cases [31–33]
SLC30A10	Normal [34]	NA	NA
SLC39A14	NA	NA	NA
NPC	Abnormal (10)	NA	Cortical and thalamic hypometabolism [36,36]
Chediak-Higashi Syndrome	Abnormal [37]	NA	NA
Ataxia Telangiectasia	Normal	Abnormal in some cases [38]	Reduced uptake in cerebellum [39]
Cerebrotendinous Xantomatosis	Abnormal [40]	NA	Cerebellar hypometabolism [41]
DYT-GCH1/ATP1A3	Normal [42–44]	NA	GCH1: increases metabolism in dorsal midbrain, cerebellar vermis, and SMA, decreased in the putamen, lateral premotor and motor cortical regions [45–47]
DYT- PRKRA	Abnormal [48]	NA	NA
Lubag TAF1	Abnormal [49]	Abnormal [50]	Reduced uptake in basal ganglia [51]
SCAs	Abnormal in some cases [52–54]	Abnormal [55]	Reduced uptake in cerebellum (also in basal ganglia in SCA3) [56]
FXTAS	Abnormal in some cases [57]	Abnormal [58,59]	Cortical hypometabolism [60]
POLG/TWNK	Abnormal [61]	NA	Thalamic, parietal and occipital hypometabolism [62]
SPG genes	Abnormal in some cases [63]	NA	Cortical hypometabolism [64]

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