ATP13A2 variants in early-onset Parkinson's disease patients and controls

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Four genes responsible for recessively inherited forms of Parkinson's disease (PD) have been identified, including the recently discovered ATP13A2 (PARK9) gene. Our objective was to investigate the role of this gene in a large cohort of PD patients and controls. We extensively screened all 29 exons of the ATP13A2 coding region in 112 patients with early-onset PD (EOPD; <40 years) of mostly European ethnic origin and of 55 controls. We identified four carriers (3.6%) of novel single heterozygous ATP13A2 missense changes that were absent in controls. Interestingly, the carrier of one of these variants also harbored two mutations in the Parkin gene. None of the carriers had atypical features previously described in patients with two mutated ATP13A2 alleles (Kufor-Rakeb syndrome). Our data suggest that two mutated ATP13A2 alleles are not a common

cause of PD. Although heterozygous variants are present in a considerable number of patients, they are - based on this relatively small sample -