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DCTN1 mutation analysis in families with progressive supranuclear palsy-like phenotype

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We recently read the article by Caroppo et al.[1] with interest, and we congratulate them for their excellent work. Caroppo et al.[1] analyzed 19 families in which affected individuals presented with atypical parkinsonism and a progressive supranuclear palsy (PSP)-like phenotype. They also identified a DCTN1G71E mutation in four of the affected patients; all were from a single French family. The mutation has already been reported previously in another French family.[2]

Interestingly, Cases II-7 and III-5 only partially fulfilled the current clinical diagnostic criteria for possible PSP.[3] Therefore, it remains to be seen whether classic PSP cases are indeed carriers of this specific gene mutation. On the other hand, two of their other cases, Cases III-7 and III-12, displayed three out of the four cardinal clinical features of Perry syndrome, which were also accompanied by frontal lobe signs. Perhaps, it would be better to diagnose these cases as having atypical Perry syndrome instead of behavioral variant of frontotemporal dementia because frontal lobe signs have previously been described in Perry syndrome.[4] Additionally, three of the cases that were diagnosed with Parkinson's disease also had a rapid clinical course (I-1, II-2, III-3) and additional features such as hypoventilation, which is strongly suggestive of atypical parkinsonism. Finally, in several previously described cases with Perry syndrome, levodopa was helpful, but the daily dose was above 1000mg, so it would be interesting to know the dose of levodopa for Case III-3.[5] It also would be interesting to find out if there is a founder effect between their family and the previously reported French family with members carrying the same mutation.

We compliment Caroppo et al. for the interesting manuscript and the description of an additional family with this very rare disease. Their manuscript sheds additional light on phenotypic presentation of Perry syndrome.

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Disclosures

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