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Cerebellar and thalamic degeneration in spinocerebellar ataxia type 10. The devil is in the details

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The spinocerebellar ataxias and SCA10 in particular, are rare diseases. We are glad that Arruda et al. are reporting new information regarding the brain atrophy in SCA10 [1,2]. Their results support our findings [3], but also highlight the phenotypic heterogeneity between the two main subsets of SCA10: 'Mexican' and 'Brazilian' SCA10. The Brazilian type (without seizures) is mostly found in southern Brazil, while the rest of the cases found in several South American countries corresponds to the Mexican subtype characterized by the presence of seizures [4]. At the genetic level, the repeat composition of the expanded allele is the main determinant of the risk of seizures; ATCCT interruptions between the ATTCT and ATCCC tracts increase the likelihood of epilepsy greatly [5]. Morphologically, our study of Mexican SCA10 patients showed that significant thalamic degeneration was strongly correlated with seizures [3]. It is therefore not surprising that the SCA10 patients in the report from Arruda et al. do not show major thalamic degeneration, which is consistent with their lack of epilepsy.

Future studies of Mexican SCA10 patients from families without seizures and Brazilian SCA10 patients with seizures are warranted to further delineate the role of the composition of the mutant allele and the resulting patterns of brain damage in determining the disease subphenotypes. Collaborative efforts will be key to achieve these goals. This work was funded by UNAM grant DGAPA-PAPIIT No. IN220019, CONA-CYT A1-S-10669 to JFR

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