CRANIAL NERVE ATROPHY IS A DISTINCTIVE FEATURE OF RFC1-RELATED DISORDER.

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Background: Differential diagnosis between RFC1-related disorder (RFC1), spinocerebellar ataxias (SCA) and multiple system atrophy cerebellar type (MSA-C) may be challenging. Thinning of cranial nerves V (CNV) and VIII (CNVIII) has been suggested as a distinctive RFC1 sign, but its disease-specificity is unclear. **Aims**: To assess the usefulness of CNV and CNVIII atrophy as a neuroradiological sign to differentiate RFC1 from SCA and MSA-C. **Methods**: Forty-eight patients with late-onset ataxia were enrolled, including 16 with RFC1, 23 with SCAs (types 2,3 and 6) and 9 with MSA-C. Ten healthy controls were also assessed. We acquired axial T2-weighted MRI with thin slices for qualitative assessment of CNV and CNVIII. A blinded neuroradiologist assessed MRIs. Frequencies of combined and isolated atrophy of nerves V and VIII were compared between groups using Fisher exact test. **Results**: Atrophy of CNV was significantly more frequent in the RFC1 group when compared to SCA, MSA and control group ($p=0.003$, $p<0.001$, $p<0.001$; respectively). Atrophy of CNVIII was significantly more frequent in the RFC1 group when compared to MSA and control group ($p= 0.002$, $p=0.003$; respectively), but not in comparison to SCA group (62.5% vs 39.1%, $p=0.2$). In an exploratory sub-analysis assessing each SCA subtype, simultaneous atrophy of both nerves was also more frequent in the RFC1 group than in SCA2 ($p=0.026$), SCA3 ($p=0.009$) and SCA6 ($p=0.005$) subgroups. **Conclusion**: CNV and CNVIII thinning is a specific neuroradiological sign of RFC1 easily detected by dedicated MRI sequences. **Keywords**: RFC1, CANVAS, Ataxia.