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Clinical scales and vestibulo-ocular reflex as biomarkers of pre-clinical stages in Machado-Joseph disease/Spinocerebellar ataxia type 3 (BIGPRO Study)

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BACKGROUND: There is scarce knowledge about natural history of presymptomatic stages of Spinocerebellar Ataxia Type 3/Machado-Joseph Disease (SCA3/MJD). BIGPRO is a longitudinal study aiming to validate biomarkers for disease progression in SCA3/MJD since pre-clinical periods (bigpro.webnode.com). Vestibulo-ocular reflex (VOR) alterations could be one of them. AIM: to report baseline findings obtained from clinical scales and VOR parameters registered by video-oculography. METHODS: Baseline data were collected from 30 symptomatic and 59 at 50% risk SCA3/MJD subjects. Genetic tests performed in at risk subjects were double-blind. For presymptomatic carriers, time left until the onset of gait ataxia was estimated by their CAGexp and was called "time to onset"; they were classified as far from (AFF) or near (AN) (4 years or less) the predicted age at onset (AO). Time to/time after onset (TtoAfterOnset) was the dimension of time to all SCA3/MJD carriers. SARA, SCAFI, NESSCA and INAScount were obtained. VOR was measured by video-oculography (EyeSeeCam): the average gain observed at 60ms from the start of the head impulse (VOR60) was considered. Bonferroni corrections was used; different letters mean pairwise significances. RESULTS: CAGexp and TtoAfterOnset of 30 symptomatic, 13 AN and 24 AFF were respectively 75.40 (3.06), 77.00 (3.19) and 74.21 (2.38) repeats (ns); 4.5 (0 to 8), -4.85 (-6 to -4) and -14.46 (-29 to -7) years. Clinical scales and VOR of symptomatics, AN, AFF and controls were all significantly different between groups ($p<0.05$): NESSCA 13.63 (7-23)a, 6.85 (2-13)b, 2.75 (0-9)c and 1.77 (0-5)c; SARA 8.02 (3-16)a, 1.27 (0-2.5)b, 0.625 (0-2.5)b and 0.5 (0-1.5)b; ICARS 22.27 (8-47)a, 6 (2-13)b, 2.88 (0-9)c and 1.68 (0-7)c; INAScount 5.57 (2-11)a, 3.54 (0-7)b, 1.63 (0-5)c and 1.14 (0-4)c; SCAFI -0.77 (0.76)a, 0.10 (0.35)b, 0.38 (0.41)c and 0.67 (0.40)c; and VOR60 0.69 (0.20)a, 0.87 (0.19)b, 1.02 (0.07)c and 1.04 (0.09)c, respectively. TtoAfterOnset of the 37 presymptomatic carriers correlated ($r=0.443$ to 0.627) with ICARS, NESSCA, VOR and INAScount. CONCLUSION: VOR60, NESSCA and ICARS were the best candidate biomarkers for the presymptomatic period in SCA3/MJD. Our longitudinal observation will try to confirm these findings.