

of N83, N376, N1056 and N1129 for GlcNAc-1-phosphotransferase activity suggest that the  $\alpha$ - and  $\beta$ -subunits form a catalytically active complex, in which N-linked oligosaccharides of the stealth domain 1 and 2 are most likely part of the UDP-GlcNAc binding site.

### eP3204

#### **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.

### eP3205

#### **Clinical scales and eye movements show changes in time since pre-clinical stages in Machado-Joseph Disease/Spinocerebellar Ataxia type 3 (Bigpro Study)**

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**BACKGROUND:** BIGPRO is a longitudinal study aiming to validate biomarkers for disease progression in Spinocerebellar Ataxia Type 3/Machado-Joseph Disease (SCA3/MJD) since pre-clinical periods (bigpro.webnode.com). Causal treatment is not available yet and there is scarce knowledge about natural history of presymptomatic stages. **AIM:** to report baseline findings obtained from clinical scales and eye movement recordings with video-oculography, including saccades, pursuit, gaze-evoked nystagmus and central nystagmus. **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, INAScount were obtained. Eye movements were measured by video-oculography (EyeSeeCam) as: reflex vertical saccade velocity (RVS), gains of vertical and horizontal pursuits, slow-phase velocity of gaze-evoked (SPV-GE) and central nystagmus (SPV-C). Bonferroni correction 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 results were described elsewhere, in other abstract. RVS, SPV-GE and SPV-C of symptomatics, AN, AFF and controls were also significantly different: 237,59b (52,0), 241,71b (57,87), 314,88c (59,0) and 336,35c (52,67); 1.50(1.30)a, 0.57(0.46)b, 0.27(0.33)c and 0.18(0.24)c; and 0.41(0.40)a, 0.29(0.26)b, 0.11(0.08)b and 0.10(0.11)b, respectively. Time to onset of 37 presymptomatic carriers showed decreasing  $r$  (from 0.627 to 0.363) with ICARS, NESSCA, INAScount, horizontal pursuit, RVS and SPV-C, respectively. **CONCLUSION:** These results suggest that reflex vertical saccade velocity is the best candidate as biomarker among eye movement parameters for the pre-symptomatic period in SCA3/MJD – similar to NESSCA and ICARS. Longitudinal observations will deepen these observations and perhaps confirm these findings.

### eP3212

#### **Saúde ambiental: avaliação do ambiente em regiões sob influência de usina termelétrica movida a carvão**

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O carvão mineral é o combustível fóssil mais utilizado para a geração de energia elétrica. O carvão tem estrutura química complexa, que consiste em uma mistura composta por carbono, hidrogênio, enxofre, oxigênio, nitrogênio que estão associados a outros elementos rochosos e minerais inorgânicos. A exposição ao carvão e seus efeitos genotóxicos e mutagênicos vem sendo estudados com diferentes enfoques e em diferentes organismos. O objetivo deste trabalho foi avaliar a instabilidade genômica e alterações epigenéticas de diferentes populações que vivem no entorno da região de exploração e queima do carvão, relacionando com o ambiente e tendo em conta a direção preferencial de ventos, considerando a fonte poluidora, de queima de carvão. Para este estudo foram coletadas amostras de solo em todas as cidades a serem avaliadas (Aceguá, Bagé, Candiota e Pinheiro Machado), no período de inverno e verão. Foi realizada a identificação e a quantificação dos elementos inorgânicos pelo método PIXE. Para realização deste estudo foi cultivada a linhagem celular de V79 (pulmão de hamster chinês), estas foram expostas aos extratos das amostras de