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Audiological and vestibular pathologies in subjects harbouring the mtDNA mutation A3243G

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Presentation Abstract

Title: E-P06.01 - Audiological and vestibular pathologies in subjects harbouring the mtDNA mutation A3243G

Keywords: A3243G; Audiological; Vestibular

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Abstract: **Introduction:** The mitochondrial DNA point mutation A3243G is known to express mainly two syndromes: Maternally Inherited Diabetes and Deafness (MIDD) and Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-like episodes (MELAS). Research has found Sensorineural Hearing loss (SNHL) to be the most frequent symptom in subjects bearing the A3243G mutation. Dysfunction of the vestibular organs is, however, scarcely investigated. With this study, we aim to examine the impact the A3243G mutation has on the inner ear. This was done by performing extensive vestibular and audiological examinations. **Method:** Eight subjects with blood verified A3243G mutation underwent thorough audiological and vestibular examinations: Tone and speech-audiometry, video Head Impulse Test (v-HIT) of all six semi-circular canals (SCC), ocular- and cervical-Vestibular Evoked Myogenic Potential (VEMP), and otoneurological examination. Subjective symptoms were evaluated with the Dizziness Handicap Inventory (DHI) questionnaire. **Results:** SNHL was found in seven subjects, one suffered a conductive hearing loss, combined mean PTA₄ of 58,4 dB. Speech Discrimination (n=7) ranged from 24% to 100%, mean at 74%. v-HIT (n=42) found pathology in nine lateral SCCs, five posterior SCCs and one anterior SCC, three measurements were inconclusive. All (n=14) o-VEMPs were absent, nine c-VEMPs were absent and two were inconclusive. Through the DHI, six subjects reported none to mild dizziness, one reported moderate, and one severe dizziness. **Conclusion:** Our population showed pathology from all the audiological and vestibular end organs. Our findings indicated that the pathology was situated within the end organs, and not within the superior- and/or inferior vestibular nor cochlear nerves. **Grants:** None

Presentation Time: Sunday, May 28, 2017, 9:00 AM - 5:45 PM