

DOCTORIAL DISSERTATION IN PSYCHOLOGY

Abstract

Winblad, S. (2006). *Myotonic dystrophy type 1. Cognition, personality and emotion*
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Myotonic dystrophy type 1 (DM1) is a multi-system disease caused by an unstable CTG triplet repeat expansion on chromosome 19. The syndrome primarily affects skeletal muscle but anomalies in brain, cognition and personality are frequently present. The main aim of this thesis was to explore cognition, personality and facial emotion recognition ability in patients with classical DM1 (onset age 10 or later). Analysis of associations was performed between behavioural domains, CTG repeat expansion size in blood lymphocytes and cerebrospinal fluid biomarkers. **Study I** used the Temperament and Character Inventory (TCI) to map personality in DM1. Findings indicated aberrations regarding Harm avoidance, Persistence, Self Directedness and Cooperativeness. Furthermore signs of a personality disorder were found in 20% of the DM1 patients. CTG repeat expansion size did not correlate with any of the personality dimensions or the presence/absence of personality disorder. The aims of **Study II** were to investigate facial emotion recognition ability and to explore genetic-, neuropsychological, and personality correlates of this ability. Results revealed an impaired recognition of facial emotion expression and a significant negative correlation was found between total score on a forced choice emotion recognition task and CTG repeat size. Furthermore, specific cognitive functions (vocabulary, visuoconstructive ability and speed) and personality dimensions (Reward dependence and Cooperativeness) correlated with scores on the forced choice emotion recognition task. In **Study III** cognitive abilities were explored. Patients with DM1 scored worse in comparison to normative collectives on tests measuring executive, arithmetic, attention, speed and visuospatial abilities. Furthermore it was found that larger CTG repeat size was associated with lower results on most tests associated with these abilities but also overall cognitive ability. **Study IV** examined cerebrospinal fluid content associated with neuronal degeneration and amyloidogenesis (tau and β -amyloid) and their associations with cognitive abilities. As compared to gender and age matched controls β -amyloid was significantly decreased, while levels of CSF-tau were increased. Levels of β -amyloid correlated significantly with WAIS-R performance IQ. In sum, this thesis shows that DM1 is a disorder associated with neurocognitive dysfunction, anomalies in personality and a reduced ability to recognize facial emotion. The results also indicate that blood CTG repeat expansion size is associated with cognitive and facial emotion recognition abilities. Overall, these findings underline the importance of considering behavioural anomalies when seeing patients with DM1 in clinical practise and the need for further research on neurocognitive aspects of this disease.

Keywords: Myotonic dystrophy, CTG-repeat expansion, Personality, Facial emotion recognition ability, Neuropsychology, β -amyloid, Tau

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ISBN 91-628-6978-7
ISSN 1101-718X
SRN GU/PSYK/AVH-181-SE
