Laboratory approach to the diagnosis of narcoplepsy

Thelen MHM, Soons JWPH, Leuverman JG, Steinmann RJAA, De Groen JHM*
Laboratory for Clinical Chemistry, St-Annaziekenhuis, Geldrop, *Centre for sleep/wake disorders, Kempenhaeghe, Heeze

Purpose: Narcolepsy is a REM sleep disorder of as yet unknown aetiology, marked by severe daytime sleepiness and cataplexy. In order to support the cumbersome clinical diagnosis of narcolepsy we wanted to evaluate a laboratory approach that combines sensitive screening with specific confirmation.
Method: Patients suffering from a sleep disorder and suspected for narcolepsy were analysed by polysomnography during 2 nights which is the established golden standard for this diagnosis. The same patients were tested for HLA DQB1*0602, which has a prevalence of 24% in the general population. Patients were also tested for hypocretin levels in CSF.
Results: Using polysomnography as a reference, 40 of 72 suspected patients indeed had narcolepsy. PCR analysis for HLA-DQB1*0602 showed a sensitivity and specificity of 97% and 70% respectively. This establishes HLA-DQB1*0602 as a good marker for screening. However, it lacks specificity to confirm the diagnosis. Using 30% of the value found in controls as a cut-off CSF hypocretin had a sensitivity of 91% and a specificity of 97.5% for narcolepsy. This makes CSF hypocretin the ideal marker to confirm the diagnosis.
Discussion: Hypocretin in CSF is a good discriminator between narcolepsy and other sleep disorders. When patients are screened for HLA-DQB1*0602 first, spinal taps can be confined to those patients with positive HLA-DQB1*0602 screening.

Marc H.M. Thelen, Laboratory for Clinical Chemistry, St-Annaziekenhuis, PO BOX 90, 5660 AB, Geldrop, Netherlands, t +31-(0)40-2864866, m.thelen@st-anna.nl

poster Neuroscience 1