P36

The role of [123I]-FP-CIT in differential diagnosis between CANVAS and MSA-C: an useful tool in clinical practice?

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Introduction: CANVAS (Cerebellar Ataxia, Neuropathy, Vestibulare Areflexia Syndrome) is a rare adult-onset neurodegenerative disorder usually due to a pathogenic AAGGG expansion in the RFC1 gene (cr. 4). One-third of patients present with dysautonomia, which can lead to a Multisystem atrophy type C (MSA-C)-like phenotype [1].

Objective: We studied nine Italian late-onset ataxia patients, observed at the Neurological Unit of the Federico II University, harboring the homozygous AAGGG expansion in RFC1 to clarify whether [123I]-FP-CIT is a valuable tool to differentiate CANVAS and MSA-C patients.

Methods: Genomic DNA from peripheral blood leukocytes were tested by standard PCR with primers flanking the CANVAS locus. Clinical features were estimated using the Scale for the Assessment and Rating of Ataxia (SARA) and investigations included MRI, DATscan with I-123-Ioflupane, FDG-PET.

Results: Cerebellar atrophy was present in all but one patients and it was associated with pons atrophy in one. [123I]-FP-CIT was abnormal, with a mild to moderate decrease of tracer uptake, in three tested patients without parkinsonian features. There was no statistically relevant difference in [123I]-FP-CIT findings between CANVAS and MSA-C patients.

Conclusion: An abnormal [123I]-FP-CIT does not seem to contribute to differential diagnosis between CANVAS and MSA-C: genetic testing for CANVAS expansions is still a fundamental diagnostic tool in patients with late-onset ataxia.

References:

[1] Cortese A, Tozza S, Yau WY, Rossi S, Beecroft SJ, Jaunmuktane Z, Dyer Z,Ravenscroft G, Lamont PJ, Mossman S, Chancellor A, Maisonobe T, Pereon Y,Cauquil C, Colnaghi S, Mallucci G, CurroR, Tomaselli PJ, Thomas-Black G, SullivanR, Efthymiou S, Rossor AM, Laurá M, Pipis M, Horga A, Polke J, Kaski D, HorvathR, Chinnery PF, Marques W, Tassorelli C, Devigili G, Leonardis L, Wood NW,Bronstein A, Giunti P, Züchner S, Stojkovic T, Laing N, Roxburgh RH, Houlden H,Reilly MM.(2020) Cerebellar ataxia, neuropathy, vestibular areflexia syndromedue to RFC1 repeat expansion.Brain. 143:480-490.