Sporadic progressive ataxia with palatal tremor (PAPT): clinical and neuroimaging clues to the diagnosis

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sPAPT is a very rare neurodegenerative disorder characterized by high signal and hypertrophy, on T2-weighted brain MRI images, of the inferior olivary nuclei (ION)

79-year-old male with hypertension, hyperlipidemia, type II diabetes and coronary heart disease; no family history of neurological diseases.
2 years ago subacute gait unsteadiness. First Brain MRI: subacute ischemic lesion in the left internal capsule. Afterwards slowly progressive gait ataxia, more recently complicated by speech disturbances and mild dysphagia.
Neurological examination: saccadic pursuit and slightly hypometric saccades without nystagmus, a slow and irregular speech, a constant 1-2 Hz palatal tremor, standing and gait ataxia, a slightly positive Romberg's sign and frenage at limb coordination tests. Deep tendon reflexes were diminished, no pyramidal or extrapyramidal signs
Blood tests (including serum lactic acid, vitamin B12, folate and thyroid hormones): normal.
Brain MRI : cerebellar atrophy and simmetric increased signal and hypertrophy of the ION only in T2-weighted sequences .
Brain PET/CT (after the administration of 185 Mbq of 18F-FDG): hypermetabolism of olivary nuclei bilaterally.
Search for antibodies for immune-mediated forms of cerebellar ataxia (TPO, GAD, transglutamine, antigliadine, antionconeuronal and other neural Ig autoantibodies): negative.
Genetic tests (AD-SCA 1,2, 3, 6, 7, 17 , FXTAS, POLG1 and FRDA): negative.
Neuropsychological assessment: mild attention defects
EMG/ENG: sensory-motor axonal polyneuropathy,
Electrooculography: normal amplitude but reduced duration and velocity for saccades; normal gain, phase and asymmetry but reduced frequency values of smooth pursuit

Conclusion: Our findings support the occurrence of functional iperactivity underlying the peculiar brain MRI changes in PAPT and their correlation with the appearance of palatal tremor. This report confirms that neuroimaging studies together with a careful neurological examination, including palatal motility , allow to address the correct diagnosis of this rare neurodegenerative condition.

Figure 1

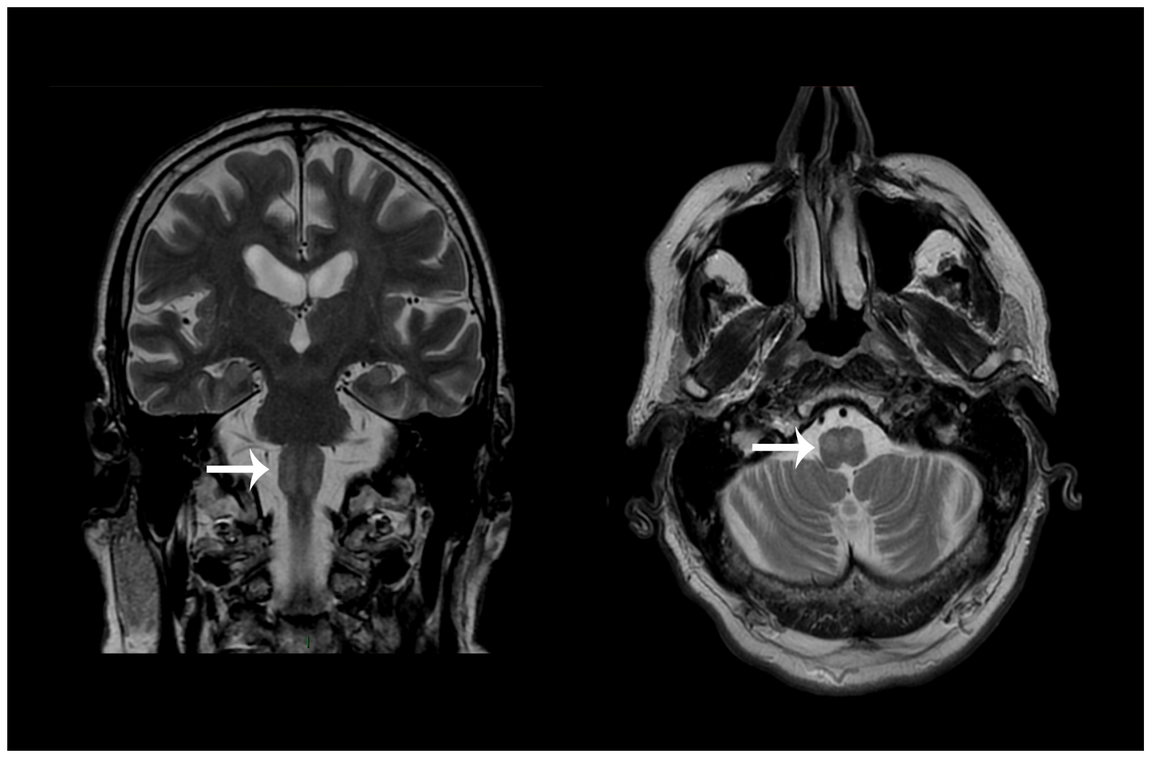

Figure 2

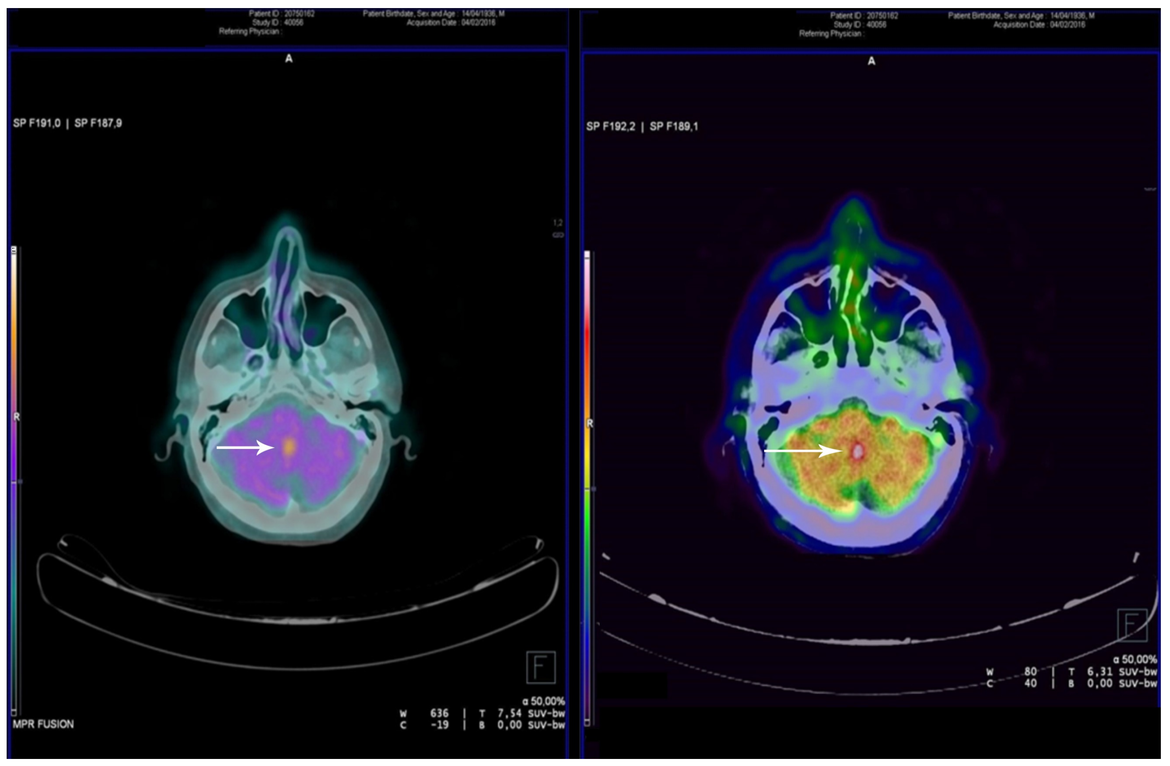

Fig 1: Axial and coronal sections of T2-weighted brain MRI, showing high signal and hypertrophy of the inferior olives (arrows) and cerebellar atrophy

Fig.2: Brain [18F] FDG-PET reveals hypermetabolism of the olivary nuclei (arrows)

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