CASE STUDY

Cerebellar Ataxia With Neuropathy and Bilateral Vestibular Areflexia Syndrome (CANVAS) in an Imbalance Patient∗

El síndrome de ataxia cerebelosa con neuropatía y arreflexia vestibular bilateral como explicación de una inestabilidad progresiva incapacitante

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Clinical Case

A woman of 63 years of age, with a history of high blood pressure, type 2 diabetes mellitus and hypothyroidism and no family history of cerebellar disease. Her gait started to become progressively unstable 5 years ago (at age 53) and she has required walking aids for 2 years and experiences difficulties with speaking. She does not report experiencing vertigo.

At the time the diagnosis was made, the patient presented abolition of the visuo-vestibular ocular reflex with clear signs of ”doll’s eye” and tonal audiometry showed a curve suggestive of symmetrical presbycusis in line with the age of the patient.

Complementary neuro-otological tests showed bilateral gain reduction on video-head impulse test (v-HIT) (Otometrics®) (0.36 mean right gain, 0.27 mean left gain) with evident refixation saccades demonstrating bilateral vestibular loss (Fig. 1). The oculomotor study performed with Ulmer’s video-nystagmography (Synapsis®) showed alteration of slow and optokinetic tracking with preservation of saccadic movement (Fig. 1). A study using the posturographic NedSVE (IBV®) system showed an overall PDC of 64%, a sensory organisation test of 36% with a mixed vestibular and somatosensory loss pattern and stability limits of 37%.

The neurological study showed a heel-toe gait with widened base gait, bilateral heel-knee dismetry, scanning speech, cephalic tremor of negation, hypoaesthesia and distal areflexia in the lower limbs.

Neuropsychologic study of the extremities showed a severe sensory axonal polyneuropathy predominating in the lower limbs with an absence of sensory evoked potentials.

Radiological study with brain MRI revealed atrophy of the cerebellar vermis and both hemispheres (Fig. 2).

Blood, biochemical, vitamin B12 and folic acid tests were normal, TSH: 7180 (0.340–5600); T4: 0.1 (0.60–1.60); HIV, Lues, Borrelia and Brucella negative, anti-amphiphysin...
antibodies, Hu, Yo, CV2, Ma2 negative. Molecular study of spinocerebellar ataxia (SCA) for SCA1, SCA 2, SCA 3, SCA 6, SAC 7 and DRPLA for Friedrich’s ataxia were negative.

Discussion

Cerebellar ataxia with neuropathy and bilateral vestibular areflexia syndrome has been newly described.\textsuperscript{1,2} Authors of English-speaking literature have proposed the name CANVAS which is the acronym for the description Cerebellar Ataxia, Neuropathy and Vestibular Areflexia Syndrome. This clinical picture can be distinguished from other forms of bilateral vestibulopathy with cerebellar atrophy which have been described since 1991.\textsuperscript{3} The same group of authors\textsuperscript{1} who describe the current syndrome, described a similar one (CABV syndrome) characterised by the absence of visuo-vestibular ocular reflex combined with cerebellar involvement, but their description at that time did not cover the association between vestibular areflexia-cerebellar ataxia and sensory neuropathy as a component of a differentiated clinical picture. The clinical picture is based on the doll’s eye sign which represents the combination of the failure of the 3 compensatory systems of eye movement, i.e., a change in optokinetic reflex, in slow tracking and in vestibulo-ocular reflex. Added to this is the presence of idiopathic non-hereditary cerebellar ataxia which is clearly differentiated from other more aggressive forms (multiple system atrophy – cerebellar type or Creutzfeldt-Jakob disease)\textsuperscript{4} and, finally, the existence of sensory axonal neuropathy with the absence of sensory action potentials.

The clinical picture was described in 27 patients with equal gender distribution, of late onset (age range from 33 to 71) and a mean of 11 years evolution of the disease (3–38 years). The clinical characteristic in most patients is a lack of balance. This is the reason that the authors who describe the syndrome\textsuperscript{1,2} flag up that CANVAS should be suspected clinically in patients with a lack of balance due to severe ataxia and sensory axonal neuropathy with an absence of sensory action potentials. Other frequent signs among these patients are slow saccadic tracking, gaze-evoked nystagmus, ataxic gait, scanning speech and

Figure 1  Left: study of the vestibular ocular reflex with vHIT (Otometrics\textsuperscript{®}) which reveals low gains and the appearance of open and covert saccades. Right: disorder of slow and optokinetic tracking (Ullmer-Synapsis\textsuperscript{®}).

Figure 2  Brain MRI showing cerebellar atrophy.
dismetry. The authors describing the syndrome suggest that this disease is a late-onset recessive disorder and, as described from post-mortem study of the temporal bone of one of these patients, is characterised by a vestibular ganglionopathy, with a loss of Purkinje cells at cerebellar level and sensory neuronopathy. Rehabilitation treatment is not very effective due to the alteration of the 3 main balance control systems.

References


