CASE REPORT

The first case of Hereditary Motor-Sensory Neuropathy and Cerebellar Atrophy (HMSNCA) diagnosed in Europe in a patient referred for early onset dementia evaluation

Konstantina G. Yiannopoulou[1], Stavroula Papaikonomou[2], Anastasia Yiannaki[1], Evanthia Diakoumopoulou[2], Kleanthis Karydakis[1], Nikolaos Katsilambros[2]

1. Neurological Department, Laiko Hospital, Athens, Greece
2. Department of Propaedeutic Medicine, Athens University Medical School, Laiko Hospital, Athens, Greece

ABSTRACT

Introduction
Hereditary Motor - Sensory Neuropathy (HMSN) associated with early onset dementia, cerebellar atrophy and hypoalbuminemia has been recognized as a distinct form of hereditary multisystem neuronal degeneration named HMSNCA (HSMN associated with cerebellar atrophy) and has been reported exclusively in Japanese families.

Case presentation
We report a case that fullfills the criteria for HMSN associated with cerebellar atrophy (HMSNCA), being the first case reported in Europe. The patient was a caucasian 63 years old woman coming from Netherlands and referred for early onset of dementia. The reported symptoms were confusion, progressive memory and orientation impairment, gait disturbance, tremor, peripheral numbness and amyotrophy of the lower limbs. Neurological examination revealed moderate dementia, signs of peripheral sensory-motor polyneuropathy and cerebellar signs. Laboratory tests revealed transient hyponatremia and hypoalbuminemia. The neurophysiological examination was indicative of chronic demyelinating sensory-motor polyneuropathy. Brain MRI demonstrated significant cortical, subcortical and cerebellar atrophy.

Keywords: *dementia, hereditary motor and sensory neuropathies, hypoalbuminemia, cerebellar disorder*

INTRODUCTION

Families with hereditary sensorimotor neuropathy, cerebellar atrophy, dementia and hypoalbuminemia have been increasingly reported in Japan.[1-4] Their common features are the autosomal recessive inheritance, the early onset dementia, the cerebellar ataxia and the symptoms and signs of neuropathy. The above mentioned cases were initially considered as a variant form of Friedreich ataxia. However, subsequent clinical studies, including five Japanese families, have led Fukuhara et al to propose a new disease entity, named Hereditary Motor and Sensory Neuropathy associated with Cerebellar Atrophy (HMSNCA).[5] Neuropathological and genetic features of HMSNCA are still unknown, and the nosological position of HMSNCA has not been settled yet. We report the first European case consistent with HMSNCA.

Address correspondence to: G. Yiannopoulou,
Vas. Tsounia 12A, Maroussi 11 526, Athens, Greece
Tel.: +30-2107456592 / 6932 281946, Fax.: +30-210 7712686
Email: ekati2@otenet.gr

CASE REPORT

Our patient was a 63 year- old woman coming from Netherlands. On November 2005 the woman was referred to Hospital because of severe confusion, generalized muscle weakness and urinary incontinence of 20 days duration. According to her birth history, she was full-term and normally delivered. At around age 25, mild tremor appeared in the hands while writing or eating. Several years later, muscle weakness and sensory disturbance of the limbs were also noticed. These symptoms slowly progressed and at around age 40 the patient developed severe gait disturbance. Since then, a memory and orientation impairment was gradually developed, it was first observed at age 58, and progressed eventually to the current condition. On the neurological examination, the woman was moderately demented, with a Mini-Mental-State Examination (MMSE) score of 18. Her extraocular movements were moderately limited in all directions with horizontal gaze-evoked nystagmus. The extremities showed combined claw(ulnar nerve impairment) and ape hands (median nerve impairment) and "stork-leg" appearance (muscle mass loss in the lower part of distal extremities). Distal dominant muscle atrophy and weakness, sensory disturbance of glove and stocking type, generalized areflexia and intentional tremor of the upper limbs were also observed.

Routine blood studies showed severe hyponatremia (Na=120mmol/l), a decreased total protein (5,9g/dl) and albumin (3,2g/dl) and increased total cholesterol (283mg/dl) and triglycerides (242mg/dl) in the serum. Monoclonal protein in serum or urine was not detected. Further investigation resulted in normal findings for thyroid, liver and renal function, vitamin B_1, B_6, B_{12}, and E, amino acid analyses in blood and urine, chest roentgenogram and electrocardiogram (ECG).