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Spinocerebellar Ataxia Type 3 in Italy: Time to Change Mind

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In a review on the global epidemiology of hereditary ataxias published in this journal in 2014, it has been reported that spinocerebellar ataxia type 3 (SCA3) is considered to be absent in Italy [1]. Similar results were also published in a prevalence study in the province of Padua in 2004 [2]. However, isolated Italian SCA3 families have been previously reported [3, 4]. Here we report the cases of 3 additional Italian SCA3 patients. Patient 1 is a 54-year-old man with progressive unsteady gait since age 47. At neurological examination, he showed an ataxic gait but could walk unaided. Other cerebellar signs included moderate scanning speech, intentional tremor, mild dysmetria and dysdiadochokinesia as well as bilateral fixation nystagmus and saccadic pursuit. Facial fasciculations were also noted. NCS/EMG evaluation showed mildly reduced cMAP amplitudes with normal NCVs, consistent with motor axonal neuropathy. Neither pyramidal and extrapyramidal signs nor mental retardation was detected. Brain MRI showed moderate pancerebellar atrophy. His 50-year-old affected sister presented with similar clinical features since age 45. Interestingly, bilateral exophathalmus, a typical sign of SCA3, was observed in both patients. Direct mutation analysis for CAG expansion in *ATXN3* [5] showed a heterozygous CAG expansion (CAG number 73 ± 3 in both patients). The family is of Umbrian ancestry with autosomal dominant pattern of transmission. Patient 3 is a 65-year-old man with unsteady gait since age 55. He showed gait ataxia, horizontal and vertical ophthalmoparesis, mild dysarthria, dysphagia, dysmetria and dysdiadochokinesia. Except for a bilateral extensor plantar, no pyramidal signs were noted. NCS/EMG evaluation revealed axonal neuropathy and MRI showed mild cerebellar atrophy. He harboured a heterozygous CAG expansion in *ATXN3* (CAG number >55).

In conclusion, our report supports the fact that there is ongoing common practice of SCA3 testing even in Italy.

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