PostScript

LETTERS

Pigmentary macular dystrophy in spinocerebellar ataxia type 1

Spinocerebellar ataxia type 1 (SCA1) is an autosomal-dominant, late-onset, slowly progressive disorder, primarily characterised by a gradual loss of motor coordination, resulting from dysfunction and degeneration of the cerebellum and its connecting pathways.^{1,2} This disease is caused by expansion of a CAG trinucleotide repeat within the *SCA1* gene, which encodes a protein called ataxin-1.^{1,2} The first and most typical neurological symptom of SCA1 is cerebellar ataxia. Additional symptoms comprise the slowing of saccades, ophthalmoplegia, dysarthria, pyramidal sign and involuntary movement.^{1,2} As regards ophthalmological abnormalities of SCA1, decreased visual acuity owing to optic atrophy, attenuation of oscillatory potentials of electroretinogram and enlargement of corneal endothelial cells have been reported.³ Hence, pigmentary macular dystrophy (PMD) has not been recognised as a clinical feature of SCA1, although it is well established as a major symptom of spinocerebellar ataxia type 7 (SCA7), which is caused by expansion of a CAG trinucleotide repeat within the *SCA7* gene.^{4,5} Here, we report a patient with SCA1 who developed visual impairment due to PMD indistinguishable from that of SCA7.

The patient is a 56-year-old Japanese man, whose father died of stroke at the age of 49 years and whose mother died of gastric cancer at the age of 70 years. He has a 54-year-old sister, who was diagnosed with spinocerebellar degeneration elsewhere, although details are not known. He has no other brothers or sisters. At the age of 45 years, he noticed an unsteadiness of gait and slurred speech, both of which progressed gradually. He began to use a wheelchair by the age of 53 years. Neurological examination at the age of 56 years showed a slowing of saccadic eye movements, scanning speech, facial dyskinesia, bilateral limb ataxia and pyramidal signs. Brain magnetic resonance imaging showed atrophy of the pons and cerebellum.

The patient began to experience visual impairment at the age of 51 years. Ophthalmological examination at the age of 56 years showed that his best-corrected visual acuity was 0.2 OD and 0.1 OS. Intraocular pressures were normal and his media were clear. Fundus examination showed mild atrophy of the macular retinal pigment epithelium, with very small clumps of pigment in both eyes (fig 1). Foveal reflex was lost and a coarse granular appearance was noted in the macular region, but choroidal neovascularisation was absent. No signs of inflammation were noticed either in the anterior segment or in the retina. A relative central scotoma was detected in both eyes by Goldmann perimetry. Ishihara and panel D15 colour vision tests showed an inability to distinguish any colours. Single-flash electroretinography showed negative β wave pattern and mild attenuation of the oscillatory potentials. Phototropic electroretinography also showed mild attenuation of

Figure 1 Photograph of the patient's left fundus. Coarse granular appearance with pale areas of pigmentary atrophy in the macular region was noted. The small clumps of pigment in pale areas were observed by fundus examination with slit-lamp biomicroscopy. No abnormalities were observed outside the macular region. Similar findings were present in the right fundus.

each wave. On the basis of these findings, we diagnosed for his visual disturbance as PMD.

As both his fundoscopic and electroretinographic findings closely resembled those reported in SCA7 PMD,⁴ we suspected that the patient might have SCA7. We carried out genetic diagnosis with the informed consent of the patient. We obtained a blood sample from the patient, extracted genomic DNA from the peripheral leucocytes and then analysed the CAG repeat-containing fragment in the *SCA7* locus, using previously described conditions.⁵ Unexpectedly, the patient had homo normal alleles with 14 CAG repeats, excluding the diagnosis of SCA7. Next, we analysed the *SCA1* locus using similar methods.¹ The patient had a normal allele with 26 CAG repeats and an expanded allele with 51 CAG repeats. The sequences were as follows: (CAG)₁₃CAT CAG CAT (CAG)₁₀ in the normal allele and (CAG)₅₁ in the expanded allele. We diagnosed the patient's illness as SCA1 on the basis of these results.

The SCA1 patient reported here developed PMD indistinguishable from that of SCA7. At present, we do not know if the PMD observed in our patient is an unusual phenotype of SCA1 or a rare coincidental complication. For instance, an interesting possibility is that a causative gene of hereditary PMD might be located in the vicinity of the *SCA1* locus and mutated in our patient. In either case, our findings indicate that SCA1, in addition to SCA7, should also be considered in the differential diagnosis of hereditary spinocerebellar degeneration complicated by PMD.

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Reinfection with Lyme borreliosis presenting as a painful polyradiculopathy: Bannwarth's, Beevor's and Borrelia

Lyme disease is caused by a tick-borne spirochaete, *Borrelia burgdorferi*. Infection may be asymptomatic, may cause only erythema migrans (spreading from the site of the tick bite) or may cause disseminated disease affecting many organs, including the nervous system. Infection with *B burgdorferi* is transmitted by ixodid ticks, which are found in many parts of the UK. Their activity is seasonal, with peaks in summer and early autumn. The risk of human infection mirrors tick activity, with peak periods in summer and early autumn, although patients may present with later-stage manifestations throughout the year.

A 53-year-old man presented in September 2004 with a 3-week history of pain in the left flank and lumbar region, associated with hypersensitivity in the left lower abdomen; 5 days before admission, the right side also became affected. Constipation and hesitancy of micturition, with preserved sensation, had been present for 3 days. No weakness in the legs, fever or rash was observed. The patient was a non-smoker and drank <30 units of alcohol a week. He was regularly exposed to the risk of tick bite owing to the area of residence and recreational activities, but he denied recent tick bites. Six years earlier, the