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Macular degeneration as a common cause of visual loss in spinocerebellar ataxia type 1 (SCA1) patients.

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BACKGROUND: Spinocerebellar ataxia type 1 (SCA1) caused by pathogenic CAG repeat expansion in the ATXN1 is characterized by loss of vision with little fundus abnormalities in some patients. Recently, macular degeneration has been reported to account for the visual symptoms in sporadic cases.
MATERIALS AND METHODS: Five consecutive patients diagnosed as SCA1 with supporting genetical evidence were newly referred to ophthalmology department from neurology unit. They underwent ocular examination to assess visual acuity and the structural integrity of the macula using optical coherent tomography (OCT). Full-field and multifocal electroretinogram (ERG) were recorded in some patients. Genetic testing was done by a polymerase chain reaction-based method.
RESULTS: Fundus examinations revealed normal optic disc and macula appearance. However, four out of five patients had foveal thinning by OCT. This included three patients who showed reduced visual acuity. Among the three, multifocal ERG was performed in two, which showed reduced amplitudes in the localized foveal area. Full-field ERG showed normal responses in all five patients assessed. Only one patient had normal visual function and normal macular structure.
CONCLUSIONS: Macular degeneration with subtle funduscopic alterations, sometimes mimicking occult macular dystrophy, is an important cause of visual loss in SCA1 patients, which could be reliably detected with OCT and multifocal ERGs.

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