cerebellar syndrome. These patients usually have cerebellar cortical atrophy. Autosomal dominant cerebellar ataxia resembles idiopathic cerebellar ataxia in many respects. Like idiopathic cerebellar ataxia, autosomal dominant cerebellar ataxia is a late onset disorder that may present as a pure cerebellar disorder or in combination with non-cerebellar symptoms. Neuropathological findings in idiopathic cerebellar ataxia and autosomal dominant cerebellar ataxia are often indistinguishable. These phenomenological similarities have prompted the hypothesis that at least some cases of cerebellar ataxia, from spontaneous mutation of a gene responsible for autosomal dominant cerebellar ataxia, from non-paternity, or falsely appear sporadic due to incomplete pedigree data. The recent discovery of an expanded trinucleotide repeat mutation in the SCA1 region in American families with autosomal dominant cerebellar ataxia offers the opportunity to search for this mutation in isolated patients with sporadic cerebellar ataxia by the polymerase chain reaction.

We have analysed DNA from 61 patients with idiopathic cerebellar ataxia with a mean age at onset of 15 years and mean disease duration of 9.2 (SD 47) years. All patients satisfied the following diagnostic criteria: (a) progressive, otherwise unexplained ataxia, and (b) family history without evidence of heredity or consanguinity of parents. All diagnoses were made after exclusion of possible symptomatic causes (alcoholism, other toxic causes, malignancy, hypothyroidism, vitamin deficiency, inflammation, metabolic, or vascular causes). Twenty five patients had a pure cerebellar syndrome (idiopathic cerebellar ataxia-C), whereas 36 had additional non-cerebellar symptoms (idiopathic cerebellar ataxia-P). Of these, 17 fulfilled the clinical criteria for probable multiple system atrophy. Polymerase chain reaction analysis was performed, as described by Orr et al. For comparison, DNA from 144 patients with autosomal dominant cerebellar ataxia was analysed. As a positive control, DNA from families with autosomal dominant cerebellar ataxia families derived from Siberia was included, in which the SCA1 expansion has been demonstrated and sequenced. A diagnosis of SCA1 heterozygocity was made in five out of 19 German families with autosomal dominant cerebellar ataxia for which the family history was medically documented. This corresponds to a prevalence of SCA1 in German families with autosomal dominant cerebellar ataxia of about 25%. By contrast, the SCA1 expansion was found in none of the 61 cases of idiopathic cerebellar ataxia. The present results argue against the hypothesis that the number of cases of idiopathic cerebellar ataxia are due to a trinucleotide expansion at the SCA1 locus. The importance of our failure to show a mutation of the SCA1 gene in patients with idiopathic cerebellar ataxia is underlined by the finding that the SCA1 mutation is present in a considerable proportion of German families with autosomal dominant cerebellar ataxia. This data does not exclude the possibility that some cases of idiopathic cerebellar ataxia are due to spontaneous mutations at other gene loci associated with autosomal dominant cerebellar ataxia. Linkage studies currently being done on some of these families show that there are at least two other gene loci (SCA2 and SCA3) for autosomal dominant cerebellar ataxia. The hypothesis that idiopathic cerebellar ataxia is due to a mutation at one of these loci cannot be tested at present because SCA2 and SCA3 genes are not identified. It is unlikely that a recessive gene defect accounts for many of idiopathic cerebellar ataxia because recessive inheritance of a late onset ataxic disorder has been documented only very rarely. It therefore seems likely that most cases of idiopathic cerebellar ataxia are non-inherited, an hypothesis supported by a recent study suggesting that idiopathic cerebellar ataxia in monzygous triplets may be more common in autosomal dominant cerebellar ataxia. Pathological and neuroradiological studies show that there is frequent spinal involvement in autosomal dominant cerebellar ataxia but not in idiopathic cerebellar ataxia. Finally, postmortem studies have shown that the presence of oligodendroglial intracytoplasmatic inclusions in the brains of patients with sporadic olivopontocerebellar atrophy, but not in the brains of patients with dominantly inherited olivopontocerebellar atrophy. 

KLOCKGETHER K, BURK J B, SCHULZ J, DICHEENS Department of Neurology, University of Tubingen, Hoppe-Seyer-Straße 3, D-72076 Tübingen, Germany

Correspondence to: Dr T Klockgether, Department of Neurology, University of Tubingen, Hoppe-Seyer-Straße 3, D-72076 Tübingen, Germany

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Atypical presentation of vascular events in pituitary tumours: "non-apoplectic" pituitary apoplexy

Sudden disabling headache, visual deterioration, or impaired conscious level (sometimes in the context of a known pituitary adenoma under medical treatment) is the classical presentation accompanying acute haemorrhage or infarction of the pituitary gland. We have recently managed three cases in which radiologically and pathologically confirmed vascular events in pituitary tumours presented with ophthalmoplegia in the absence of changes in either visual fields or acuity and in which headache was a minor symptom. This atypical mode of presentation may confound accurate diagnosis and delay appropriate treatment. Surgery for this condition is associated with good functional recovery and low morbidity in most cases. It is therefore important that such atypical presentations are recognised early so that neurosurgical advice can be sought.

Case 1, a 48 year old man of Syrian origin, developed mild headaches and gastrointestinal upset while visiting Syria. He was initially treated for bacterial dysentery and his symptoms subsided. Within 48 hours, however, he had developed double vision. At the time of neurological assessment he seemed well, alert, and was free of headache. On examination there were complete left third and sixth nerve palsies though there was no apparent impairment of fourth or fifth nerve function. Visual acuity, visual fields, and fundoscopy were all normal. No signs of meningism were present and the rest of the neurological examination was normal. Brain MRI showed an intrasellar mass compressing the left cavernous sinus and displacing the optic chiasm (figure). Magnetic resonance angiography performed at the same time excluded an aneurysm. At transphenoidal hypophysectomy infarcted pituitary tissue that was under some tension was evacuated; there was a rim of apparently normal tissue evident at the periphery of the lesion. After the operation there was a rapid improvement in the diplopia, which has subsequently completely resolved. Histopathological examination showed infarcted pituitary tissue; there was insufficient viable tumour to allow for immunohistochemistry.

Case 2, a 52 year old man, developed a sudden drooping of his left eyelid accompanied by a mild headache. There had been no alteration in his vision and there was no history of endocrine disorder. Examination showed ptosis and proptosis of the right eye with complete ophthalmoplegia and pupillary involvement. Fundoscopy was normal. Corrected visual acuity was 6/9 on the right and 6/5 on the left; the visual fields were full. Magnetic resonance imaging showed a sellar mass of mixed high signal with suprasellar extension to the optic chiasm which was clearly compressing the right cavernous sinus and displacing the right internal carotid artery. The appearances were typical of tumour infarction with haemorrhage. At transphenoidal hypophysectomy a large quantity of overtly necrotic pituitary tumour was removed. Histology confirmed necrosis and haemorrhage. Immediately after operation the ocular movements began to improve. At the time of discharge the fourth and sixth nerve palsies had fully recovered but the third nerve palsy remained evident.

Case 3, a 78 year old woman, originally presented with ophthalmoplegia due to a pituitary adenoma. This had almost completely resolved 18 months after transphenoidal hypophysectomy when she developed diplopia of a few days duration accompanied by drooping of the left eyelid. The patient complained of a mild discomfort behind the left eye but no headache. She was alert and lucid. A pupil sparing left third nerve palsy and also a partial left sixth
Life threatening epilepsy in a child.

T Kuroiwa, H Morita, H Tanabe and T Ohta

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