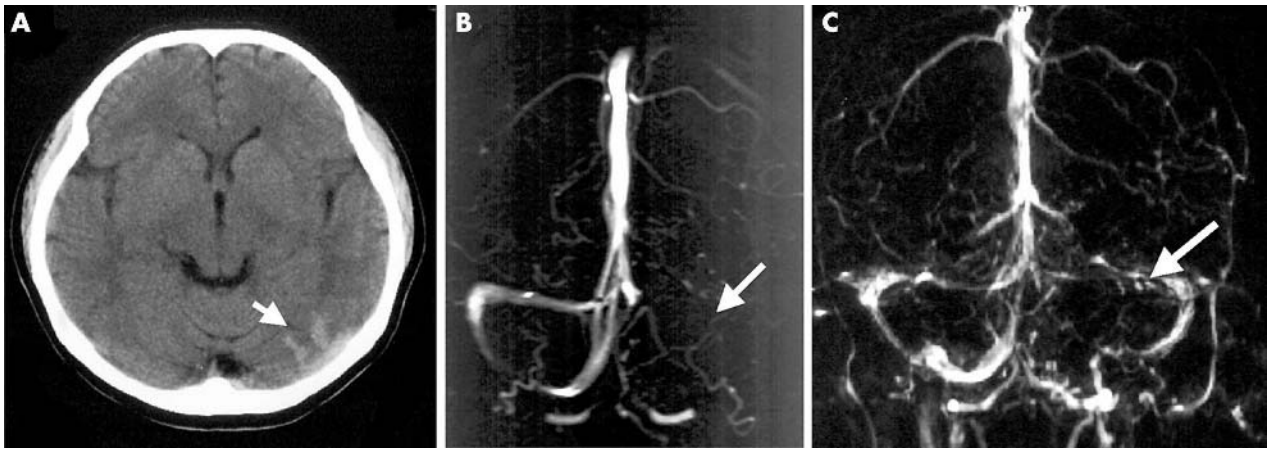

Figure 1 (A) Brain CT shows a high density lesion (arrow) in the left temporo-occipital lobe. (B) MRV (ECG gated two dimensional time of flight) shows a lack of flow in the left lateral sinus (arrow). (C) In the follow up MRV, the left lateral sinus (arrow) is recanalised with a residual stenosis.

In conclusion, physicians should be aware that Cushing's syndrome is a possible cause of cerebral sinus thrombosis. Plasma levels of factor VIII and VWF may play an important role in the hypercoagulable state in hypercortisolism.

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Spinal muscular atrophy, Dandy-Walker complex, and cataracts in two siblings: a new entity?

Lower motor neurone involvement is the main feature of several neurological disorders, including the various forms of spinal muscular atrophy (SMA). A distinct form of SMA is characterised by predominantly distal weakness and atrophy of the limbs.[1] Various combinations of SMA with neural and extraneural defects, mainly pontocerebellar hypoplasia, have also been reported.[2]

We report a combination of distal SMA with Dandy-Walker complex and anterior polar cataracts in two brothers.

The patients were aged 25 and 23 years. Their parents, who originated from the same area of Greece, were unrelated and asymptomatic. Since the age of 10 years, both brothers presented with progressively deteriorating symmetrical distal muscle weakness and atrophy of the lower limbs, which affected mainly the anterior tibialis and peroneal muscles and, to a lesser degree, the gastrocnemius, resulting in an almost ''stoke-like'' appearance of the legs. Bilateral anterior polar cataracts had been diagnosed in both patients at the age of 9–11 months. Additional findings of the neurological examination in both patients were slight muscle strength reduction in both hands and forearms and decreased tendon reflexes in the upper and lower limbs, while the Achilles' tendon reflexes could not be elicited. The plantar responses were normal. No sensory, cerebellar, or cognitive impairment was found. Dysmorphic features were not observed. The general physical examination was normal in both patients.

Extensive haematological, biochemical, and immunological investigation of both patients, including levels of creatine kinase, prolactin, hexosaminidase A, anti-GM_1 and antisulphatide antibodies, cortisol, thyroid hormones, vitamin B_{12} and folic acid, immunoglobulin and lipoprotein electrophoresis, and cerebrospinal fluid examination, was normal.

The electrophysiological examination revealed findings compatible with anterior horns involvement in both patients. Specifically, the electromyogram showed chronic active denervation of distal muscles, with large amplitude motor unit potentials and presence of polyphasic potentials and spontaneous activity (fibrillations and positive waves), more pronounced in the lower limbs. Nerve conduction studies showed normal motor and sensory conduction velocities, with normal amplitudes, latencies, and F waves. No conduction blocks were recorded. Electrophysiological investigation of both parents was normal. Both patients refused consent for muscle biopsy.

Magnetic resonance imaging revealed the presence of Dandy-Walker complex in both patients. There was enlargement of the cisterna magna, with slight hypoplasia of the vermis and slight elevation of the tentorium (fig 1). No supratentorial or brainstem abnormalities were observed. The magnetic resonance imaging of the spine was normal in both brothers, as were visual and brainstem evoked responses. Ophthalmological examination confirmed the presence of anterior polar cataracts in both patients.

The karyotype was normal in both patients. Molecular genetic analysis for mutations in the survival motor neurone (*SMN*; exon 7 and 8 deletions), neuronal apoptosis inhibitory protein (*NAIP*; exon 5 and 6 deletions), and androgen receptor genes was negative.

Discussion

Our patients were two brothers with almost identical clinical and laboratory findings. One of the main features was the involvement of the anterior horn cells, which was compatible with distal SMA, according to published criteria.[1] Additional features were Dandy-Walker complex and bilateral anterior polar cataracts.

There are several reports of SMA with additional features (SMA plus), among them pontocerebellar hypoplasia. These cases, however, are characterised by proximal muscles involvement, early presentation with profound floppiness at birth, mental retardation, and cerebellar signs.[3] There are also rare reports of recessive distal SMA with additional features: diaphragmatic paralysis or pyramidal signs.[4] None of these cases of proximal or distal SMA plus has been linked to chromosome 5q.

Familial cases of Dandy-Walker complex are not uncommon; however, the combination of the disorder with SMA seems to be quite unusual. The aforementioned severe cases of pontocerebellar hypoplasia in SMA plus clearly constitute a different nosological entity.

The coexistence of early onset cataracts with neuromuscular disorders is also unusual. Apart from the well known occurrence of cataracts in myotonic dystrophy, there are some reports of cataracts in combination with spastic paraparesis, spinocerebellar degeneration or neuropathy, and facial dysmorphism.[5] There is also a report of familial congenital cataracts and Dandy-Walker anomaly with lissencephaly.

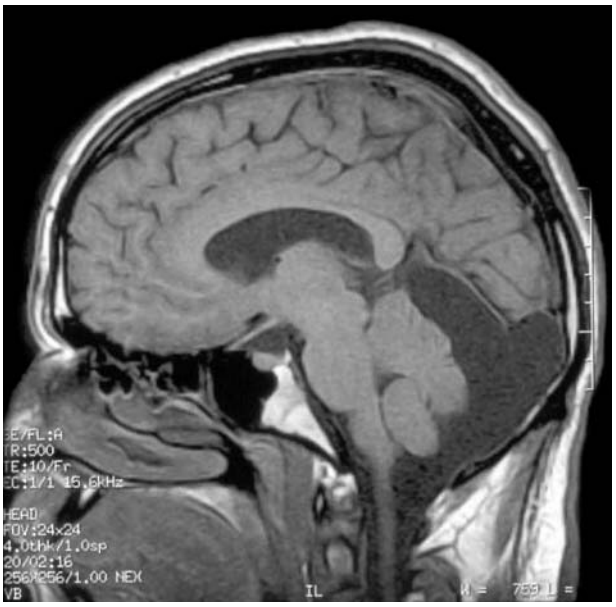

Figure 1 Brain magnetic resonance imaging of the older sibling. T1 and T2 weighted sagittal images, showing enlargement of the cisterna magna and slight elevation of the tentorium.

We were not able to locate in the literature any reports of distal SMA in combination with any form of Dandy-Walker variant and/or congenital cataracts.

In summary, our cases represent an unusual combination in distal SMA. This combination does not seem to fit in any of the already described syndromes and could be the result of pleiotropy, contiguous gene syndrome, or chance. The fact that the patients were first degree relatives and presented with identical phenotypes is a strong indication that the disorder is genetically determined. With the available information on the genetics of the main features of our patients, contiguous gene syndrome appears unlikely. We were not able to locate a genetic defect, a not altogether unexpected result, as most recessive distal SMA families remain to be genetically determined.[2] Future investigation of similar cases should include genetic studies relevant to all three main features of the disorder.

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BOOK REVIEWS

The A–Z of neurological practice. A guide to clinical neurology

Edited by Roger A Baker, Neil Scolding, Dominic Rowe, Andrew J Larner. Published by Cambridge University Press, 2004, £45.00 (paperback), pp 936. ISBN 0-52162-960-8

This pocket sized book consists of a comprehensive series of entries from A to Z, each one describing a specific aspect of neurology. The authors provide overviews of major disease groups (eg, headache, epilepsy) as well as more detailed descriptions of specific disease categories (eg, SUNCT syndrome, gelastic epilepsy) throughout 936 pages. The entries are organised in a structured way and usually include information on pathophysiology, clinical features, investigations and diagnosis, differential diagnosis, and treatment and prognosis. Some literature is quoted and extensive cross references to other entries are provided.

This is a very useful reference book for everyone who works in clinical neurology or related areas. It can also be used by general physicians who need some fast and succinct information on neurological issues. For obvious reasons this book cannot replace a textbook. The overviews of the major disease groups provide only the basic information, and the entries are of limited value for differential diagnosis and therapy. The main advantage of this ''guide to clinical neurology'' is that it provides relevant and up-to-date information on each neurological topic in a readable and accessible manner. This is of particular interest if the treating neurologist or generalist is confronted by one of the numerous rare neurological disorders and/or syndromes. This goal is also achieved by the myriad of entries and cross references. In summary, we can recommend this reference book as a useful supplement to the traditional textbooks in the neurologist's bookshelf.

J C Möller, W H Oertel

Cranial neuroimaging and clinical neuroanatomy

Edited by Hans-Joachim Kretschmann, Wolfgang Weinwich. Published by Thieme, 2003, €199 (hardback), pp 451. ISBN 3-13-672603-0

Already a well established reference, this third edition of *Cranial Neuroimaging and Clinical Neuroanatomy* was significantly updated, thereby offering a more comprehensive approach to neuroanatomy than did any of the previous editions.

This book is divided into 10 chapters and an atlas. The introduction gives an overview of the scope of this book but also provides very useful information on ''basics'' such as the various imaging planes, their historical evolution, and their definitions. The next chapter then briefly describes the various neuroimaging techniques illustrating the useful approaches to the imaging of the various areas of the head and neck.

This is then followed by the atlas, which is really at the heart of this book, as is emphasised by the subtitle. The atlas is subdivided into four sections—one section for each of the three planes and a fourth section for the posterior fossa. This edition retains in all of these sections the excellent line drawings of previous editions, which were obtained from gross anatomic slices. These are now complemented by new, large sized, state of the art T1 and T2 weighted MR images in all three planes as well as CT images in the axial plane. These additions increase significantly the practical utility of this atlas, which is enhanced by the fact that each of the four sections has a coloured margin of its own.

The topography of the neurocranium, the craniocervical junction, and the pharynx are succinctly discussed in the following two chapters covering among others the skull, the CSF spaces, the vascular territories and the subdivisions of the brain. Of particular interest are the newly introduced three dimensional reconstructions of the vascular tree, as well as the detailed schemes of the vascular supply of the posterior fossa.

The well illustrated chapter of the Neurofunctional Systems relates the anatomy with the physiology thereby underlining the clinical utility of this book. This is finally followed by the last chapter offering a succinct overview on neurotransmitters and neuromodulators.

In summary, this an excellent companion for students and medical trainees that will help them both in their initial as well as in their more advanced stages in getting a better command of the complex but very seductive world of neuroanatomy.

T A Yousry