secondary causes of intracranial hypertension can be life-threatening. Review of the imaging studies by an experienced neuro-radiologist is of paramount importance. Low threshold for repeat imaging studies should be maintained especially when symptoms do not respond to treatment or change features. Neurosurgical advice, when sought early, is likely to secure good outcome.

REFERENCES

Spontaneous Intracranial Hypotension Syndrome in a Patient With Marfan Syndrome and Autosomal Dominant Polycystic Kidney Disease

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Intracranial hypotension is typically manifested by orthostatic headache. The most frequent underlying factor is cerebrospinal fluid leakage. It has been suggested that dural structural weakness in some connective tissue diseases may be responsible for dural tears and diverticula and consequently leakage. We present a case of spontaneous intracranial hypotension associated with Marfan syndrome and autosomal dominant polycystic kidney disease. The patient was treated successfully with epidural autologous blood patch. Dural involvements of these hereditary connective tissue diseases are also discussed.

Key words: spontaneous intracranial hypotension, dural leak, epidural blood patch, polycystic kidney disease, Marfan syndrome

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INTRODUCTION

Spontaneous intracranial hypotension (SIH) is a rare cause of chronic headache in adults.\(^1\) We present a case of this condition occurring in a patient with both Marfan syndrome and autosomal dominant polycystic kidney disease (ADPKD) and emphasize dural involvement of underlying connective tissue disorder.

CASE REPORT

A 32-year-old woman with a chronic headache was referred to our neuroradiology department for cranial magnetic resonance angiography (MRA) examination. She had been diagnosed with polycystic kidney 3 years earlier (Fig. 1). Presence of intracranial aneurysm was the proposed cause of the headache. Her cranial MRA examination was normal with no detection of intracranial aneurysm. During questioning of headache profile, a dramatic postural component, in which the pain was virtually eliminated by lying flat but returned on standing up, was defined. Typical Marfan skeletal habitus, which had been neglected previously, was now recognized. At this point, a consultation with the genetics department was requested. On examination, myopia and astigmatism were detected as minor criteria of Marfan syndrome. In addition, 5 major (pectus excavatum, scoliosis >20\(^\circ\), wrist and thumb signs, pes planus, upper segment/lower segment ratio <0.86) and 2 minor (joint hypermobility, high-arched palate with crowded teeth) criteria of musculoskeletal system were detected (Fig. 2). Family history findings were identified (scoliosis and protrusio acetabuli in father, aortic valve insufficiency in uncle, and cardiac failure in grandmother). According to the Ghent diagnostic criteria,\(^2\) the diagnosis of Marfan syndrome was made. In contrast enhanced cranial magnetic resonance (MR), minimal dural thickening was present, but no subdural effusion or prominent brain sagging was noted (Fig. 3). Intrathecal gadolinium-enhanced MR cisternography was performed to detect the exact localization and morphology of possible dural tear. MR cisternography was performed at the level of L4-L5 with 22-guage spinal needle. There was no spontaneous drainage of cerebrospinal fluid (CSF), confirming that the CSF pressure was below atmospheric pressure. We administered 0.5 mL gadopentate dimeglumine (Magnevist, Schering, Berlin, Germany) with 4.5 mL isotonic sodium chloride intrathecally. Axial, coronal, and sagittal fat-saturated T1-weighted images were obtained at the level of lumbar, dorsal, and cervical regions 30 minutes after the injection. Loss of dural sac integrity and dural leakages, contrast material extravasation into epidural area, and paravertebral region were detected on MR cisternography at lumbar levels on the right side (Fig. 4). We also noticed multiple meningeal cysts at the dorsal area (Fig. 5). Because her complaints did not regress despite conservative treatment for 6 weeks, epidural blood patch was applied with 35 mL blood. Two days later, the headache was fully recovered and she was able to carry out her daily activities. Three months after the treatment, there were no complaints or neurological findings.

Fig 1.—Abdominal CT exam shows multiple renal cysts on both kidneys.

Fig 2.—Joint hypermobility.
DISCUSSION

In our case, clinical findings, minimal dural thickening on cranial MR exam, evidence of CSF leakage on magnetic resonance imaging myelography, low CSF opening pressure, absence of dural puncture or other cause of CSF fistula in history, and resolution of headache within 72 hours after epidural blood patching fulfill the SIH according to the International Classification of Headache Disorders, 2nd edition.3

The occurrence of headaches that worsen in the upright position among individuals who have undergone a lumbar puncture is well-known to all physicians, and the spontaneous form of intracranial hypotension has become well recognized by neurologists and neurosurgeons.1 Spontaneous CSF leaks are the most common cause of SIH. The exact cause of spontaneous spinal CSF leaks usually remains unknown, but a combination of an underlying weakness of the spinal meninges and a trivial precipitating event is generally suspected.1 The structural dural weakness predisposes the patient to the formation of fragile meningeal diverticula or simple dural rents that allow CSF to leak into the extradural space.1 The presence of an underlying, systemic, connective tissue disorder was first reported in 1994,4 and subsequent retrospective studies reported findings suggesting connective tissue disorders for 16-36% of patients with spontaneous spinal CSF leaks.5,6

Marfan syndrome is a relatively common autosomal dominant hereditary disorder of connective tissue with prominent manifestations in the skeletal, ocular, and cardiovascular systems. In classical Marfan syndrome, changes in connective tissue integrity can be explained by defects in fibrillin-1, a major component of extracellular microfibrils.7,8 The etiology of dural ectasia in Marfan syndrome is unknown, but is conjectured to be related to constitutionally weak spinal dura. Although dural ectasia is common in Marfan syndrome, there has been insufficient research on dura. The morphology of the lumbar dura in Marfan syndrome has not been described, as it has in other tissues been affected by Marfan syndrome. The association between this disease and connective tissue disease has recently been raised, especially since patients with Marfan’s syndrome have been reported to develop CSF hypovolemia.9,10

Autosomal dominant polycystic kidney disease is the most frequent inherited kidney disease with a prevalence ranging from 1 in 400 to 1 in 1000.11 Although renal cysts and renal failure are the cardinal manifestations of this disorder, ADPKD is a systemic disease with multiple extra-
renal manifestations, encompassing both cystic involvement of other organs (liver, pancreas, intracranial arachnoid) and connective tissue abnormalities (intracranial aneurysms and dolichoectasia, cardiac valvular abnormalities, dissection of the thoracic aorta, and cervicocephalic arteries).\textsuperscript{11}

The co-occurrence of ADPKD and Marfan syndrome has been discussed in a few reports in the literature. Hatelyboer et al suggest that when prominent features of connective tissue disease or vascular complications are found in ADPKD patients, alternative additional diagnoses should be considered, including the possibility of Marfan syndrome.\textsuperscript{12}

Recently, spinal meningeal cysts actually have been described in 3 adult patients with ADPKD\textsuperscript{13} (1.7% of 178 patients screened for intracranial aneurysm), whereas sporadic spinal meningeal cyst is a very rare abnormality in the general population. The cysts were multiple in 2 patients and solitary in one. The cysts were found at the thoracic level in 2 of them and at the lumbar level in the third one. The first 2 patients had a history of postural headaches. Coche et al reported a fourth patient with ADPKD who was found to harbor 7 thoracic meningeal cysts.\textsuperscript{14} Although the spinal cysts were numerous and relatively large, the patient remained asymptomatic. They concluded that in patients with ADPKD the clinical index of suspicion should also be raised for SIH because of leaking spinal meningeal diverticula.

In our case, meningeal cysts were found at the thoracic and lumbar levels. We held that the existence of both ADPKD and Marfan syndrome results in the development of these cysts. It is highly probable that the coexistence of ADPKD and Marfan syndrome causes severe dural weakness in our case. In our opinion, the rupture of meningeal cysts causes CSF leakage and manifestations of SIH. As a result, a new headache in patients with ADPKD or Marfan syndrome must raise the suspicion of the rupture of a meningeal cyst, beside rupture of intracranial aneurysm.

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