Dural ectasia and the Marfan syndrome

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The not-uncommon spinal abnormalities associated with Marfan's syndrome rarely undergird clinical problems, and neurological features accompanying such bone abnormalities are rare. In such unusual circumstances it is a widened vertebral canal that attracts attention: the substrate of such widening is dural ectasia with bone erosion, presumably due to hydraulic forces operating via the cerebrospinal fluid (CSF). When neural symptoms or findings do occur they may be related to stretching and traction mechanisms. This study of a symptomatic patient defined with reasonable clarity the abnormal anatomy, and some neurological symptom relief was achieved by attempting to alter the CSF dynamics. The relevant literature has been sampled to elucidate the condition.

Key Words: Marfan syndrome, dural ectasia, arachnodactyly, sacro-spinal meningocele

Apart from his hoped-for catholic interest in all facets of medicine, the neurosurgeon only occasionally encounters specific neurosurgical problems among patients falling into the category of the Marfan syndrome. In recent years the cardiovascular features of this autosomal-dominant heritable disorder have captured most of the attention of the clinician. These are well reviewed in the recent series of Roberts and Honig. In 1976, Finney and his colleagues reported a patient with Marfan’s syndrome who harbored a 3-cm aneurysm in the cavernous portion of the internal carotid artery. They described tortuosity of the left vertebral artery, and discussed the rare intracranial vascular defects in other conditions such as Ehlers-Danlos syndrome and pseudoxanthoma elasticum.

In his original paper of 1896, Marfan made no reference to those facets of what have come to be included in the Marfan syndrome other than the congenital deformities of the extremities which he encountered in a 5½-year-old girl with a hereditary medical history. He described the long tapering extremities and digits, the deformation of the latter, the thin and emaciated-appearing and atrophic-seeming musculature; he used the phrase “pattes d’araignée.” The term that he applied to this condition, overall, was “dolichostenomélie.” In 1902, Méry and Babonneix studied Marfan’s patient (who was by then 11½ years of age) with radiological investigations. Whereas Marfan had described no spinal column deformities, the radiological studies revealed a pronounced thoracolumbar kyphoscoliosis, and the lower ribs were described as very prominent. There was swelling of the cubital bone and a lengthening and thinning of the bones of the extremities. A facial asymmetry was also noted. These authors queried the etiology, suggesting that it might be due to syphilis and selected the term “hypochondroplasia” for the condition. Achard, also in 1902, presented the case of a young lady of 18 years with features similar to those of Marfan’s patient, but with a well-developed cranium and, by comparison, a smallness of the face. It was Achard who used the term “arachnodactyly,” a descriptive word of limited scope which has endured and displaced that used by Marfan. Achard also described the hyperlaxity of the joints of the extremities in his patient, but neither he, Marfan, nor Méry and Babonneix described any nervous system disorder. It was subsequent to these reports that the associated cardiac and ophthalmological abnormalities were emphasized; more elaborate descriptions and variations of the musculoskeletal system were added to those of Marfan, who had described the sparse subcutaneous fat, the fibrous tendon contractions, and the unusual configuration of the extremities. A recapitulation of the
various components included in the Marfan syndrome was presented in 1958 by Nelson, but the most complete and comprehensive description remains that of McKusick in 1972.

In the early years of this century, attempts were made to relate arachnodactyly to what was described as “status dysraphicus” and syringomyelia. Commonality of findings included spina bifida, kyphoscoliosis, deformities of the skull and thorax, clubbed feet, acroanosis, and reflex anomalies. However, the subsequent elements of the Marfan syndrome reveal divergencies which suggest that the similarities could be common to a variety of quite separate conditions.

In 1939, Fahey reported six patients with arachnodactyly who showed skeletal changes; they also reviewed 52 cases in which spinal deformity was mentioned as an accompaniment. Those deformities were scoliosis, kyphoscoliosis, and kyphosis in descending order of frequency. An additional series published in 1964 by Wilner and Finby of 18 patients studied radiologically led the authors to propose that the skeletal abnormalities and the body configurations were not specific for Marfan’s syndrome, but could be found in normal individuals as well as in patients with a variety of other diseases. They described mild to moderate scoliosis in nine of their 18 cases, and referred to the widening of the vertebral canal, a condition found in the patient presented here.

Case Report

This 55-year-old woman suffered, without any recognized precipitating cause, the abrupt onset of low-back pain, described as a “pulled muscle.” The pain spread into her left buttock and posterior thigh and followed a variable but worsening course over subsequent months. The lower end of her spine felt as though it were bruised and under pressure. The pain in her left buttock and left thigh also became prominent together with pain in the comparable areas on the right side, although less severe.

The patient also became aware of a numb area on the left side of her buttock, and, on straining at stool on the day of her first physician visit, her back pain was made worse. Although she described having nocturia twice a night for a number of years, which had been associated with a sense of urgency and leakage, perineal sensation was always intact except for a small area corresponding to the left-side buttock numbness.

Medical History. The patient had been told she had endometriosis and cysts on her ovaries, and had undergone several operations. In 1959, after an abdominal operation, she was advised that she had a “meningocele” in her pelvis but that it should be left alone. In 1969, she was again operated on for presumed endometriosis because of abdominal pain. In 1974, a hysterectomy was performed because of her “feeling poorly.” In 1983, she underwent surgery for a cyst or tumor “pushing on the muscles and producing a tight band sensation in her pelvis.” Her most recent abdominal procedure was in 1984, for abdominal adhesions and a hernia behind her navel. She also underwent a breast biopsy in 1962 for a benign tumor, and in 1964 an inflamed axillary node was removed.

The patient’s family history did not identify any certain consanguinity. Her father died at 73 years of age of lung cancer; her mother died at the age of 75 years of a cardiac-related event (attack or hemorrhage). She has two sisters, aged 62 and 64 years, both in good health. She has no knowledge of her maternal grandfather, whereas her maternal grandmother died in her late 50’s or early 60’s. Her paternal grandmother lived into her late 60’s. Her father was 6 ft 1 in. tall, her mother was 5 ft 2 in., and her two siblings were 5 ft 2 in and 5 ft 5 1/2 in. in height.

Presentation. The patient reported bifrontal headaches in association with her present illness, especially noted directly after standing up from a recumbent posture. She described occasional light-headedness and shaking of her right hand when writing; there were no alterations in dexterity nor other paresthesiae. She noted that mounting two flights of stairs, two steps at a time, would cause her to be short of breath and leave her with a sense of generalized weakness. She complained of occasional cardiac palpitations and had taken propranolol for the prior 8 years following a fainting episode. Her other medications included hydrochlorothiazide and a thyroid supplement.

Examination. The patient was 178.5 cm tall, with a long slender neck. Her upper segment/lower segment ratio was 0.84:1. There appeared to be some decreased elastic tissue about the mouth, and a receding lower jaw produced a mild “Andy Gump” appearance. Her fingers and hands were long and tapering as were her feet with a mild “hammer toe” configuration. She had a narrow chest and a prominent pectus excavatum. There was exquisite sensitivity and tenderness in the paramedian gluteal areas, straddling the upper portion of the gluteal fold in an area the size of the palm of one’s hand. Striae were noted across the posterior aspect of the pelvis. The only neurological deficit was a mild sensory impairment on the mesial aspect of the left buttock and upper thigh.

On April 18, 1986, the patient was admitted to the University of California at Los Angeles Hospital. At this time her blood pressure was 130/70 mm Hg. A water-soluble positive-contrast myelogram revealed a conforming thecal sac with a dramatically abnormal configuration of multiple pouches and diverticuli exiting sacral foraminae in a segmental pattern without the definition of a single meningocele sac (Fig. 1). There was a massively ectatic lumbosacral thecal malformation, with posterior scalloping of the vertebral bodies up to and including L-2 (Fig. 1 right). The myelographic study was followed by a computerized tomography (CT) scan, which showed the abnormal sac communicating...
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Fig. 1. Left and Center: Iopamidol myelograms demonstrating anterior sacral pouches of the meninges in free communication with the intraspinal subarachnoid space. Right: Iopamidol myelogram illustrating enlarged subarachnoid spaces closely applied to the posterior aspects of the scalloped vertebral bodies in the lumbar region.

with large sacral pouches, apparently connected with the subarachnoid space through wide apertures (Fig. 2).

An electrocardiogram demonstrated a sinus bradyarrhythmia and abnormal left axis deviation. An echocardiographic study disclosed normal left ventricular size and wall motion and normal left atrial size; however, there was a markedly dilated aortic root of approximately 40 to 45 mm, a dilated aortic valve anulus, and moderate aortic insufficiency.

Operation. On the day following admission, the patient underwent placement of a lumboperitoneal shunt. Approximately 6 weeks following this procedure a shunt revision was needed for a malfunctioning abdominal catheter. The assembly performed well thereafter. A postoperative intravenous urogram was normal. Immediately after the original shunting procedure the patient had complete resolution of her preoperative back and thigh pain, and lost the acute tenderness of the sacral area. She did, however, describe some posterior cranial pain which was eased by recumbency. This was noted immediately on arising and would tend to remain present while she was upright, but she considered herself much improved compared to her preoperative state. It is of note that between the initial shunt and its revision she noted a return of the sense of pressure at the end of her spine, which was again relieved after shunt revision.

Postoperative Course. Over the subsequent weeks and months the patient complained of headache which fluctuated in its location, but seemed to be in the lambdoid and cervical area, a feeling as though her head was too heavy. It was relieved when she leant her head back against something supportive and improved upon recumbency. Although this complaint slowly improved, it was replaced by low-back pain which continued for several months. As the headache improved, the back
pain became her main complaint, but she remained on her job. By March, 1987, 11 months following her initial operation, she was feeling pretty well and most of her complaints seemed to be improving. By April, 1987, however, she suffered the acute onset of cervical pain and limitation in neck mobility, which responded to C-1-2 joint anesthetization. She continued to have some gluteal ache but her low-back pain disappeared and her lower-extremity complaints were completely relieved.

In April, 1987, the patient was admitted by her personal physician to another hospital. A Holter study was normal; magnetic resonance imaging and a cine cardiac study were performed. The ascending aorta was dilated; its base measured 5.2 cm across and dilatation extended up to the aortic arch. Severe aortic insufficiency and mild mitral regurgitation were documented. No aortic dissection was seen. Overall, the patient’s neural and spine complaints were improved.

Discussion

Prior to the 1964 report by Wilner and Finby, Jefferson\(^8\) in 1955 described two cases of dilatation of the vertebral canal unassociated with any abnormalities of the nervous tissue. His first patient, a 22-year-old woman complained of a “red hot” feeling on either side of the lower thoracic spine with no other neurological symptoms or findings. Radiological studies revealed abnormalities of the lower thoracic spine, an increase in the anteroposterior diameter of the vertebral canal in the thoracolumbar region, pedicular narrowing at this same site, and interpedicular measurements at the upper limits of the Elsberg and Dyke ranges. Operative exploration with dural opening revealed a vertebral canal which was greatly distended but with normal neural elements and normal leptomeninges. His second patient, an 18-year-old woman, complained of pain in the low back and left calf in a sciatic-like distribution with slight weakness at the left ankle and some decrease of pin-prick perception on the outer side of the lower leg. Dilatation of the vertebral canal was again demonstrated, with scalloping of the posterior aspects of vertebral bodies and an increase in the interpedicular distances in the lumbar and the lower thoracic regions. Reexploration revealed the enlarged canal with normal neural elements. Jefferson reviewed other conditions associated with canal widening including diastematomyelia, cases with extensive cord abnormalities and neural disabilities, intrathoracic meningoceles, and neurofibromatosis.

It was Nelson\(^{14}\) who presented four cases of patients with the Marfan syndrome in 1958 and associated the syndrome with congenital enlargement of the spinal canal found in each. His first case demonstrated a bifid S-1 laminar arch, maldevelopment and fusion of L-1 and L-2, scoliosis, and posterior scalloping from T-12 to S-1 and (to a more limited degree) in the upper to mid-cervical area. The measurements and scalloping indicated to him a widening of the vertebral canal. His second case presented with slight kyphoscoliosis, posterior scalloping from T-10 to S-1, and canal widening. Nelson’s Case 3 disclosed a moderate thoracolumbar scoliosis with posterior scalloping from T-12 to S-3, but with interpedicular measurement within the normal range. His last case demonstrated bifid L-5 and S-1 laminae with slight posterior scalloping of L-2-4 and normal interpedicular measurements. None of these patients had any neurological abnormalities. His differential diagnostic considerations included tumors, cysts, diastematomyelia, meningocele, neurofibromatosis, and congenital enlargement of the subarachnoid space. He believed that what he was describing was the same condition as that described by Jefferson\(^8\) but Nelson, for the first time, associated these findings with the Marfan syndrome.

It would appear that, prior to the appearance of Jefferson’s report\(^8\) in 1955, abnormalities involving the meninges in association with the syndrome had not attracted attention or recognition. One exception to this is the report of Bean and Fleming\(^2\) in 1940. These authors reported a 27-year-old woman who presented in labor with a large cystic structure involving the right posterior portion of the pelvis. The patient revealed many of the characteristics of Marfan’s syndrome including the skeletal, visceral, ocular, and vascular features as well as genetic aspects in the family history. Among the findings at the time of Caesarean section was a large cyst attached to the sacral vertebrae through a foramen which admitted two fingers into the vertebral space between S-1 and S-2. The cyst was filled with a clear fluid and refilled following its initial evacuation. The sacral foramina, bilaterally, were described as larger than normal. A sacral defect was identified, and the authors, although not considering this finding a sacral meningocele, described it as an unusual cyst arising from the linings of the sacral canal, a form of spina bifida not theretofore reported in this syndrome. It is of note that, radiologically, the usual “scimitar” shape to the sacrum was not found, nor was a midline defect identified. However, no spine films were illustrated.

A case of considerable interest is that reported in 1971 by Strand and Eisenberg.\(^{18}\) This was the study of a 10-year-old boy with the physiognomy of Marfan’s syndrome demonstrating arachnodactyly, a pansystolic murmur, scoliosis, pectus excavatum, and hyperextensive joints. He presented with signs and symptoms of a spontaneous subarachnoid hemorrhage, and angiography demonstrated increased vessel tortuosity of the vertebral and carotid systems; however, there was no source of bleeding. A large pelvic cystic mass was identified and diagnosed as an anterior sacral meningocele. The dura mater was described as extremely thin. This patient demonstrated widening of the spinal canal and scalloping of the vertebral bodies, as had been described by Nelson.\(^{14}\) These authors seemed to agree...
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with Mitchell, et al.,\textsuperscript{13} who suggested that the vertebral scalloping was secondary to the loss of the normal protection which a strong intact dura mater provided (or would provide) to the posterior surface of the vertebral bodies. They further considered that, in this particular case, the anterior sacral meningocele, rather than being of developmental origin, was most likely the result of dural ectasia superimposed by an elevation in cerebrospinal fluid (CSF) pressure secondary to the subarachnoid hemorrhage. They, therefore, drew the distinction between their case and that of a true anterior sacral meningocele. It should be observed that the radiographs in this case, as reproduced, do not disclose a scimitar-shaped sacrum, but the illustrations do not allow comment as to details of the sacral bone structures.

In their 1978 review of anterior and lateral spinal meningoceles, Wilkins and Odom\textsuperscript{22} cited the various associated abnormalities encountered in this condition and reviewed all of the literature available to them. They found that, whereas meningoceles were found in connection with the cutaneous form of neurofibromatosis in 63% of the intrathoracic cases and in 45% of the lateral or anterior lumbar cases of meningoceles, this association existed in only 1.6% of anterior or lateral sacral meningoceles. Among the various other associated anomalies which they described, in addition to von Recklinghausen's neurofibromatosis, was a reference to the case of Marfan's syndrome and Eisenberg.\textsuperscript{18} In respect of meningoceles, Wilkins and Odom stated that the generalized disorder probably played a role in allowing meningeal herniation to occur, but that in the vast majority of reported cases no such underlying disorder was detected. This assessment was made against the background of 102 cases of anterior sacral meningoceles which they reviewed. In further support of this position, Villarejo, et al.,\textsuperscript{19} in 1983 brought the total number of cases of anterior sacral meningocele to 148. These authors made no reference to Marfan's syndrome as an associated condition and placed the problem into the general category of occult spinal dysraphism. They recapitulated the observation that the plain radiological studies usually revealed the well-known "scimitar-shaped" sacrum. Whether the absence of this specific deformity in the three examples of pelvic meningoceles\textsuperscript{12,18} and the current case is of diagnostic value is not assessable with so few cases. The sporadic occurrence of reports of Marfan's syndrome in association with spinal meningoceles raises the question as to whether this is a fortuitous association.

Cilluffo, et al.,\textsuperscript{4} reviewed the subject of idiopathic spinal arachnoid diverticula in 1981. They defined these as meningeal outpouchings arising near the dorsal root ganglion at a site of trauma or as a result of "congenital" meningeal abnormalities. They studied a nontraumatic group, comprising 21 patients. Of relevance to this discussion was their Case 14, which was of an 11-year-old girl with Marfan's syndrome who had complained of back pain for many years and was known to have had scoliosis for at least 9 years. She was tall and thin, with a severe kyphoscoliosis, a cardiac murmur, mitral insufficiency, scalloping and erosion of the posterior low thoracic vertebral bodies, a widened interpedicular measurement in the thoracolumbar region, and a low thoracic paraspinous mass extending through the right intervertebral foramen at T-10. An arachnoid diverticulum was encountered which communicated with the T-10 nerve root sleeve and had an extradural extension anterior and lateral to the dura extending into the chest. In contrast with the conclusions of Nelson,\textsuperscript{14} who pointed out that the integrity of the cortex of the scalloped vertebrae indicated the absence of an expanding lesion (in respect of the vertebral column defects of Marfan's syndrome), Cilluffo and colleagues expressly noted "erosion" of the posterior cortex of several low thoracic vertebrae. Gimeno\textsuperscript{7} believed that the most likely hypothesis in the production of arachnoid cysts was that a congenital defect or a weak point in the dura mater allowed the arachnoid to herniate into the epidural space; in agreement with other authors, he recognized the possible role of hydrostatic pressure (pulmonary Valsalva maneuvers) insofar as the dynamics of the CSF could influence symptoms or pathogenesis. From their radiological perspective of arachnoid cysts, Lombardi and Passerini\textsuperscript{10} wrote that extradural arachnoid cysts and meningoceles possess a number of features in common and, in theory, might well be considered manifestations of varying severity of the same dysraphic lesion. Based upon the evidence presented from these several sources, one could include any condition with attenuation of the dura mater in such a proposition.

In 1973, Weir\textsuperscript{21} reported the case of a 13-year-old boy with the characteristics of Marfan's syndrome (including ectopia lentis) who, on myelography (performed because of respiratory symptomatology), demonstrated an extradural lesion opposite a widened interpedicular space with pedicular flattening at the T10–11 level. A dilated, symmetrical distal dural tube was described with perineural sacral cysts. The contrast material did not enter the thoracic lesion, but at the time of operation an extradural cyst was identified. It was thin-walled, similar to arachnoid, and connected to the dura at the T-10 nerve root level. It exited by way of the intervertebral foramen. The cyst extended into the thorax under the parietal pleura. Its wall was composed of collagenous fibers interspersed with what appeared to be flattened arachnoid cells. The boy died a year later following aortic surgery, and, in addition to the vascular pathological changes, a large arachnoid cyst displacing the frontal and temporal lobes was found incidentally at autopsy. Unfortunately, the spinal canal was not mentioned in the postmortem findings. Weir noted in his discussion that, as has been previously discussed, the majority of lateral intrathoracic meningoceles are associated with von Recklinghausen's dis-
ease and the accompanying bone changes related to it. In these changes the cortical margins of the vertebral canal and vertebral bodies are usually normal. He observed that these findings were similar to the bone abnormalities seen in the Marfan syndrome. In Weir's case the lining of the meningocele did not resemble dura mater, and he believed that the development of the process was due to the hydraulic effects of the CSF. He raised the question: is Marfan's syndrome a mesodermal dystrophy?

Relevant to this matter is the report by Boucek, et al., published in 1981. These authors compared the profiles of the collagen cross-linking compounds in the skin and aortic tissue of patients with Marfan's syndrome with those of healthy control individuals. These comparisons revealed reduced amounts of chemically stable forms of intermolecular cross links and suggested that an attenuation of (probably) nonenzymatic steps involved in the maturation of collagen causes the defective collagen organization in the Marfan syndrome. Addressing this very matter in an editorial citing Boucek's paper, Pyeritz and McKusick indicated that it is difficult to distinguish phenotypic variation due to ex- 

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