Spinal neurological complications of achondroplasia

Results of surgical treatment

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Spinal neurological complications caused the admission of 17 patients with achondroplasia to the UCLA affiliated hospitals between 1955 and 1979. These patients constituted 41% of all achondroplastic patients admitted during that period. The spinal stenotic syndromes could be divided into three groups: Group I: thoracolumbar stenosis (10 patients); Group II: foramen magnum and upper cervical stenosis (five patients); and Group III: generalized spinal stenosis (two patients). Eleven patients underwent a total of 18 decompressive operative procedures for treatment of paraparesis, quadriparesis, sensory deficits, and sphincter dysfunction. Excellent results were obtained with patients in Group I and II, 77% of whom were ambulatory and continent postoperatively. Two patients in Group III fared less well, showing steady neurological deterioration despite multiple operative procedures.

The spectrum of spinal neurological manifestations secondary to achondroplasia is reviewed. Problems with conventional radiological studies and the potential role of computerized tomographic analysis of such patients are discussed. Recommendations for surgical technique are made. Early recognition, prompt clinical evaluation, and safe and accurate radiological analysis of spinal neurological complications of achondroplasia will allow appropriate decompressive surgical procedures to be performed. Excellent results may be anticipated in the reversal and prevention of neurological deficit secondary to achondroplasia with such an approach.

KEY WORDS • achondroplasia • spinal stenosis • computerized tomography • decompression laminectomy

ACHONDROPLASIA is a congenital, disfiguring disease of endochondral bone formation; it may be associated with catastrophic neurological complications. While achondroplasia is the most common form of rhizomelic dwarfism, with an estimated incidence of 15 per million population in the United States, it was for a long time considered an innocuous curiosity of nature, and compatible with an unlimited lifetime of normal neurological function. Early reports of the neurological complications associated with achondroplasia were provided by Dandy in 1921, and Donath and Vogl in 1925. The neurological manifestations may be divided into two general categories: 1) macrocephaly, and 2) compressive spinal cord and nerve root syndromes. The spinal neurological signs and symptoms in achondroplasia encompass a wide spectrum, ranging from mild nerve root irritation, to complete paraplegia and quadriplegia. This report summarizes the diagnostic and therapeutic experience at the UCLA affiliated hospitals with the spinal neurological complications associated with achondroplasia. The importance of early diagnosis, safe and reliable radiological studies, and appropriate surgical intervention is emphasized. Excellent results may be expected in the prevention and reversal of serious neurological deficits with such an approach.

Clinical Material

The case records of the UCLA affiliated hospitals from 1955 to 1979 were reviewed, and 41 patients with achondroplasia, ranging in age from newborn to 76 years, were identified. Of the 41 patients with achondroplasia, 17 had signs or symptoms secondary to spinal stenosis, including low-back pain, radicular
TABLE 1
Clinical summary of surgically treated achondroplastic patients with spinal complications*

<table>
<thead>
<tr>
<th>Case No.</th>
<th>Age (yrs), Sex</th>
<th>Presentation</th>
<th>Neurological Examination</th>
<th>Radiological Diagnosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>18, F</td>
<td>LBP; rapid onset paraparesis; paresthesias</td>
<td>paraparesis; sphincter paralysis; DTR's: 0</td>
<td>myelogram: complete block at T12-L1</td>
</tr>
<tr>
<td>2</td>
<td>38, M</td>
<td>LBP; claudication weakness of LE's</td>
<td>tone increased in legs; DTR increased; Babinski sign positive</td>
<td>CT: stenosis at L1-4; myelogram: block at L1-2</td>
</tr>
<tr>
<td>3</td>
<td>21, F</td>
<td>LBP; rapid paraplegia; paresthesias</td>
<td>paraplegia, sensory loss bowel, bladder: 0</td>
<td>myelogram: block at L1-2</td>
</tr>
<tr>
<td>4</td>
<td>35, F</td>
<td>LBP; claudication: paresthesias, incontinence</td>
<td>normal</td>
<td>CT: stenosis at L2-5</td>
</tr>
<tr>
<td>5</td>
<td>30, F</td>
<td>LBP; claudication</td>
<td>tone increased LE's; DTR's increased; Babinski sign; hypesthesias in LE's</td>
<td>CT: stenosis at L2-5</td>
</tr>
<tr>
<td>6</td>
<td>58, M</td>
<td>LBP; claudication</td>
<td>unknown</td>
<td>myelogram: lumbar stenosis</td>
</tr>
<tr>
<td>7</td>
<td>60</td>
<td>LBP; claudication; weakness</td>
<td>DTR's decreased in LE's</td>
<td>CT: persist. lumbar stenosis; facet hypertrophy</td>
</tr>
<tr>
<td>8</td>
<td>15, M</td>
<td>LBP; weakness</td>
<td>paraparesis</td>
<td>myelogram: L4-5 disc herniation</td>
</tr>
<tr>
<td>9</td>
<td>17</td>
<td>LBP; weakness; paresthesias</td>
<td>quadriparesis</td>
<td>unknown</td>
</tr>
<tr>
<td>10</td>
<td>18</td>
<td>LBP; urinary urgency</td>
<td>quadriparesis</td>
<td>stenosis in foramen magnum</td>
</tr>
<tr>
<td>11</td>
<td>43, M</td>
<td>LBP; involuntary spasms LE's; weakness; paresthesias</td>
<td>spasticity LE's</td>
<td>thoracic compression</td>
</tr>
<tr>
<td>12</td>
<td>52</td>
<td>weakness, paresthesias</td>
<td>ataxia of gait atrophy</td>
<td>myelogram unsuccessful</td>
</tr>
<tr>
<td>13</td>
<td>53</td>
<td>weakness; decreased ambulation</td>
<td>unknown</td>
<td>unknown</td>
</tr>
<tr>
<td>14</td>
<td>54</td>
<td>interscapular pain; decreased ambulation, involuntary spasm</td>
<td>spasticity, increased DTR; decreased sensation; Babinski sign</td>
<td>air myelogram; complete block at T6-9; spinal cord atrophy</td>
</tr>
<tr>
<td>15</td>
<td>34, F</td>
<td>weakness all extremities</td>
<td>quadriparesis</td>
<td>unknown</td>
</tr>
<tr>
<td>16</td>
<td>33, F</td>
<td>weakness all extremities; paresthesias</td>
<td>quadriparesis</td>
<td>unknown</td>
</tr>
<tr>
<td>17</td>
<td>5, M</td>
<td>weakness all extremities; paresthesias</td>
<td>quadriparesis</td>
<td>myelogram unsuccessful</td>
</tr>
</tbody>
</table>

*LBP = low-back pain; DTR = deep tendon reflexes; LE = lower extremity; CT = computerized tomography.

Dysesthesias, paresthesias, loss of tendon reflexes, neurogenic claudication, progressive paraparesis, and progressive quadriparesis. These 17 achondroplastic dwarfs with complications referable to the spinal canal could be divided into three groups: Group I included patients who had thoracolumbar spinal stenosis, with primary compression of the cauda equina or conus medullaris (10 patients); Group II patients had stenosis of the foramen magnum, with associated cervical stenosis and cervical myelopathy (five patients); and Group III patients presented with generalized spinal stenosis, with spinal cord compression at multiple levels (two patients).

Eleven of the 17 patients underwent a total of 18 operative procedures (Table 1). The age range for these patients was 5 to 62 years, and the average age at the time of operation was 30 years. All these patients were followed for at least 1 year postoperatively, except for one patient (Case 2) who was operated on in February, 1979, and another (Case 10) who died shortly after operation. The six other patients were not treated surgically, for the following reasons: 1) refusal of the patient to undergo operation; 2) minimal symptoms or signs, such as simple reflex changes in one 76-year-old man; or 3) for reasons unclear in available records.

Summary of Cases

Group I: Thoracolumbar Stenosis

Six of 10 patients with thoracolumbar stenosis (Cases 1–6) were treated surgically for progressive paraparesis (four patients) or frank paraplegia (two patients). The average age at the time of decompression thoracolumbar laminectomy was 33 years. The age range of patients at the time of operation in Group I was 18 to 58 years, and two of these patients were 21 years old or younger. The presenting symptoms in these six patients included low-back pain (six patients), intermittent claudication (four patients), paresthesias (three patients), and weakness unassociated with claudication (three patients).

All six patients benefited from decompression laminectomy, and all except one (Case 1) were able to
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### TABLE 1 (continued)*

<table>
<thead>
<tr>
<th>Operation</th>
<th>Immediate Results</th>
<th>Long-Term Results†</th>
</tr>
</thead>
<tbody>
<tr>
<td>T11–L5 lamin</td>
<td>persist, paraplegia; bowel/bladder paralysis</td>
<td>requires wheelchair; bowel/bladder normal</td>
</tr>
<tr>
<td>L2–4 lamin</td>
<td>relief of symptoms; bowel/bladder paralysis</td>
<td>unchanged from immed. result after 9 mos</td>
</tr>
<tr>
<td>T12–S1 lamin; L1–2 discectomy</td>
<td>persist. paraplegia; bowel/bladder paralysis</td>
<td>walks with braces; bowel/bladder normal</td>
</tr>
<tr>
<td>L2–S1 lamin</td>
<td>relief all symptoms</td>
<td>no recurrence</td>
</tr>
<tr>
<td>L2–S1 lamin</td>
<td>return to full ambul; urinary retention</td>
<td>no recurrence; bladder normal</td>
</tr>
<tr>
<td>L2–S1 lamin</td>
<td></td>
<td></td>
</tr>
<tr>
<td>L2–5 lamin</td>
<td>no change in symptoms</td>
<td>no change in symptoms</td>
</tr>
<tr>
<td>L2–5 lamin</td>
<td>relief of pain; numbness &amp; weakness</td>
<td>recurrence of original symptoms</td>
</tr>
<tr>
<td>L4–5 discectomy</td>
<td>relief of symptoms</td>
<td>relief of symptoms; 9-mos follow-up</td>
</tr>
<tr>
<td>L1–5 lamin subocc cran, C1–7 lamin</td>
<td>slight improvement; unable to walk</td>
<td>returned to walking with crutches</td>
</tr>
<tr>
<td>T6–L1 lamin</td>
<td>weaker; walked with crutches</td>
<td>progressive quardriparesis</td>
</tr>
<tr>
<td>T9–L2 lamin</td>
<td>relief all symptoms</td>
<td>progressive quadriparesis; died aged 19 yrs; autopsy evidence of cervical cord compression</td>
</tr>
<tr>
<td>subocc cran, C1–7 lamin</td>
<td>walked with walker</td>
<td>walked well for 8 yrs, then progressive quadriparesis</td>
</tr>
<tr>
<td>T1–3 lamin</td>
<td>walked with assistance</td>
<td>progressive quadriparesis</td>
</tr>
<tr>
<td>T4–9 lamin, T6–7 discectomy</td>
<td>no change postop</td>
<td>lost to follow-up</td>
</tr>
<tr>
<td>subocc cran</td>
<td>unknown</td>
<td></td>
</tr>
<tr>
<td>subocc cran, C1–7 lamin</td>
<td>operative death</td>
<td>walks 20 yrs postop; mild residual weakness</td>
</tr>
<tr>
<td>subocc cran, C1–2 lamin</td>
<td>minimal change</td>
<td></td>
</tr>
</tbody>
</table>

*Lamin = laminectomy; subocc cran = suboccipital craniectomy.
†Unless otherwise stated, follow-up period was 1 year or longer.

walk independently. Four of these six patients who were treated with thoracolumbar or lumbar laminectomy were completely free of symptoms or neurological deficit either immediately after operation or within 3 months of operation, including our most recent case (Case 2). Cases 1 and 3 were minimally improved or unchanged immediately after operation, and had severe persistent neurological deficit. One of these patients (Case 3) was eventually able to walk independently with bracing, and both patients were continent of urine and feces 1 year after operation.

The two patients with persistent paraplegia had suffered the precipitous onset of neurological deficit, while the remainder of patients in this group had insidious onset of symptoms. Only one of our patients was worsened by operation (Case 5); she had urinary retention postoperatively (not present preoperatively) that resolved after 3 months. The results of decompression laminectomy for those patients with insidious onset of neurological signs or symptoms (Cases 3, 4, 5, and 6) were excellent in the short term, except in Case 6; this patient had an inadequate decompression initially, and his deficit subsequently resolved with more extensive decompression. He eventually developed an intervertebral disc herniation requiring still a third operation (for discectomy), with resolution of all symptoms. An illustrative case report from this group follows.

**Case 3.** This 21-year-old right-handed woman was an achondroplastic dwarf. She had a 3-year history of tingling paresthesias and mild weakness of the lower extremities, exaggerated by standing for long periods and relieved by rest in a recumbent position. Three weeks before her admission to the hospital (approximately 1 month after the felicitous termination of a normal term pregnancy), she had the spontaneous onset of low-back pain associated with loss of bowel and bladder sensation. She was initially treated with antibiotics for a presumed urinary tract infection. Two weeks before admission, she began to complain of constant paresthesias of the lower extremities and a
progressive paraparesis. At the time of her admission, she was incontinent and had been unable to walk for 3 days.

Muscle strength was normal in the upper extremities, slightly greater than antigravity in the proximal lower extremities, and no motion could be elicited below the knees. All muscle stretch reflexes were active except the Achilles reflexes, which were absent bilaterally. There was total bowel and bladder paralysis. Plain x-ray films of the spine revealed a narrowed spinal canal throughout, particularly marked at L-5, and a severe kyphosis at the thoracolumbar junction. Myelography revealed an incomplete subarachnoid block at T-12 and a complete block at L1-2 (Fig. 1). The patient underwent an emergency T12-S1 decompression laminectomy, at which time an extruded disc was identified at L1-2 and removed. Immediately after the operation there was little change in her neurological deficits, and she was transferred to a rehabilitation facility. One year later she was continent and able to walk independently with the aid of bracing and crutches.

Group II: Foramen Magnum and Upper Cervical Stenosis

Of the five patients in Group II, three (Cases 9-11) were treated surgically with suboccipital craniectomy and cervical laminectomy (Table 1). The average age at operation was 21 years, including a 5-year-old child. The presenting symptom was a progressive quadriplegia in all three. In addition, two adults (Cases 7 and 8) also complained of paresthesias in all four extremities. One patient died due to a postoperative hematoma (Case 10). Both of the other two patients were able to walk independently after operation, and were afforded long-term improvement by the operation. A report of one of these two cases follows.

Case 11. This 5-year-old boy was achondroplastic dwarf. He was admitted to the hospital with a 3-week history of progressive quadriplegia and incontinence. His developmental landmarks had been normal, and he was an intelligent and active child. There was generalized weakness graded 4/5, more severe in the proximal than distal muscles. Neck musculature was also weak. He was able to walk unassisted only with difficulty, with a wide-based, waddling gait. His tendon reflexes were increased symmetrically, and he exhibited extensor plantar responses. Cranial nerve and sensory examinations were normal. His head circumference was 52 cm (50th percentile: 51 cm).

Roentgenographic examination of the skull revealed a small posterior fossa and a small foramen magnum (Fig. 2). Computerized tomography (CT) confirmed the presence of a small posterior fossa, and revealed a thickened occipital squama and a small fourth ventricle in normal position (Fig. 3). The CT scan also revealed mildly enlarged lateral ventricles. Posterior fossa contrast myelography was attempted via a cisternal puncture, but was unsuccessful. The patient underwent decompressive suboccipital craniectomy and C1-2 laminectomy. His postoperative condition was generally unchanged, and he was transferred to a rehabilitation facility. Steady improvement occurred in his ability to walk during the ensuing year.

Group III: Generalized Spinal Stenosis

Two patients (Cases 7 and 8) represent examples of generalized spinal stenosis in which compression of the spinal cord occurred throughout the spinal axis and included the foramen magnum. Both patients had multiple and extensive operations (Table 1). In spite of improvement after each operation, these patients deteriorated insidiously to eventual tetraparesis. A summary of the course of one of them follows.

Case 8. This 43-year-old man was an achondroplastic dwarf. He initially presented in 1963 for evaluation of low-back pain, paresthesias, weakness, and involuntary muscle spasms of both lower extremities. Motor evaluation revealed a spastic paraparesis with increased tendon reflexes and extensor plantar responses. There were no abnormalities of
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FIG. 2. Case 11. Plain skull films. Left: On the basal view, the small size of the foramen magnum is noted. Right: On the lateral view, thickened occipital squama is seen compromising the size of the posterior fossa.

bowel or bladder function. An oil myelogram revealed a complete subarachnoid block at L1–2. A T9–L2 decompression laminectomy was performed. Post-operatively the patient was relieved of all symptoms, and was discharged.

He did well for 10 years, but was readmitted in 1972 for evaluation of inability to walk and paresthesias of both upper and lower extremities. Examination at the time revealed a spastic quadripareisis with atrophy of the intrinsic musculature of both hands. The patient underwent a decompressive suboccipital craniectomy and C1–7 laminectomy, followed by a course of rehabilitation, after which he was able to ambulate with a walker. Subsequently, the patient suffered a gradual decline in his ability to walk, and 1 year later, in 1973, he was admitted to another hospital. A T1–3 laminectomy was carried out and the patient briefly regained the ability to walk between parallel bars.

In 1974, he was again admitted to UCLA Hospital for evaluation of pain in the right leg, progressive paraparesis (he was no longer able to stand), persistent interscapular pain, and paresthesias of both upper and lower extremities. He also complained of intermittent urinary incontinence. Examination at that time revealed a spastic quadripareisis with unsustained clonus throughout the upper and lower extremities, and extensor plantar responses. His strength was generally 3/5 to 4/5 in the upper extremities and 2/5 to 3/5 in the lower extremities. There was significant

FIG. 3. Case 11. Computerized tomography scan. Markedly thickened occipital squama is visualized producing constriction of the posterior fossa at and just above the foramen magnum.
atrophy of the upper extremity musculature and a
global diminution of sensation. Gas myelography
revealed atrophy of the entire spinal cord, most
marked in the cervical region, and spinal cord com-
pression at T6–9. The patient underwent T4–9 decom-
pression laminectomy and removal of a herniated disc
at T6–7. Postoperatively, the patient demonstrated
minimal improvement and was transferred to a
rehabilitation facility. He was then lost to follow-up

Discussion

Surgical Technique

Adequate surgical decompression is mandatory if
reversal of preexisting deficit and prevention of
further deterioration is to be expected. Wide lamincto-
tomy to the level of the pedicles is recommended. Use
of rongeurs with a thick foot plate is best avoided. The
high-speed drill is a useful adjunct in performance of
safe and adequate laminectomy. The short, thick
nature of the laminae makes their removal with
rongeurs particularly difficult. The high-speed drill is
useful for this maneuver, either thinning the laminae
and allowing easy removal with a delicate rongeur, or
providing complete removal, using a diamond burr for
the final portions of bone. Extrudal exploration for
extruded disc material is indicated, dependent on
myelographic findings. Only one patient in our series
showed slight deterioration neurologically in the im-
mediate postoperative period (urinary retention), and
this cleared within 3 months. One patient, who failed
to respond rapidly immediately after the operation
improved later after more extensive decompression.
One death due to a postoperative hematoma occurred
following occipital craniectomy and cervical laminecto-
my.

Bone Changes in Achondroplasia

The aberration of endochondral bone formation
present in achondroplasia causes characteristic
changes in the spine, originally described by Donath
and Vogl.7 The abnormalities occur both in the in-
dividual vertebrae and in the vertebral column as a
whole, and regularly result in spinal stenosis with
spinal cord and root compression syndromes. Nelson23
estimated that 47% of achondroplastic patients mani-
fest spinal complications. Detailed analyses of these
bone changes have been reported extensively in the
literature.1,2,3,33,34 The spinal cord, conus medullaris,
and cauda equina are of normal size and are closely
confined in the narrowed spinal canal. The shape of
the lumbar and thoracolumbar segment of the spine
becomes increasingly important as changes wrought
by aging, trauma, and postural stress result in increas-
ing thoracolumbar kyphosis, occasionally with gibbus
formation, osteoarthritic spur formation, and in-
tervertebral disc degeneration. These factors work in
contact to produce progressive constriction of a con-
genitally narrowed canal. Thus, symptoms related to
spinal cord compression in the thoracolumbar region
were believed to occur uncommonly in childhood or
eyear adulthood.18

Fifty-nine cases of impending or actual paraplegia
have been reported in achondroplastic patients; 43 of
these were treated surgically, exclusive of the present
series.1,2,3,6,10,13,18–19,21,26,30,31,33 Our experience is at
variance with the belief that transverse myelopathy or
cauda equina compression is rare before the third
decade.18 Four patients, aged 15 years or younger,
with signs or symptoms consistent with spinal cord or
root compression are described in the literature.2,9,13,25
The youngest patient described by Grossiord, et al.,13
was 12 years old when he first complained of dif-
ficulties in walking, consistent with neurogenic claudi-
cation. This patient did not undergo decompression
laminectomy until the age of 60 years. Epstein and
Malis8 reported a patient with progressive myelopathy, who was operated on at 15 years of age.
Two of our six patients with thoracolumbar or simple
lumbar stenosis were 21 years or younger at the time
of operation, and the average age of our patients, 30
years, is somewhat younger than nearly 38 years, as
reported in a recent series.18

Upper cervical myelopathy secondary to a small
foreman magnum with compression of the upper
cervical spinal cord and brain stem is a recognized,
but rarely reported, complication of achondro-
plasia.2,4,6,14,19,24,36 Foramen magnum insufficiency is a
common occurrence in achondroplastic dwarfs, and is
thought to result from the effect of the underlying
defect in endochondral bone formation on the base of
the skull.20 Lethal compression of the high cervical
cord in an achondroplastic infant was first reported by
Potter and Coverstone;44 they suggested that the high
mortality rate in achondroplastic infants is due to
compression of the brain stem at the foramen
magnum.22 This anomaly was considered the cause of
death in two achondroplastic infants reported by
Yang, et al.,46 and led them to speculate about sub-
lethal upper cervical cord injury in the etiology of the
slow motor development and hypotonicity commonly
seen in the first months of life.22 While the recently
reported cases with foramen magnum insufficiency
have included only children,19,36 we report one child
and two adults with this syndrome. We could find no
other reports in the literature of foramen magnum in-
sufficiency requiring decompression in adults. Cohen,
et al.,4 reported a case of foramen magnum stenosis in
a 5-year-old child (very similar to our Case 11), who
presented with paraparesis and difficulty in walking,
and who was treated successfully by suboccipital
craniectomy and upper cervical laminectomy. The
operative findings that they reported are similar to
those in our patient, namely, a hypertrophy of the
atlas and of the occipital bone comprising the bone
margin of the foramen magnum.

While various references to diffuse or generalized
spinal stenosis were noted in the literature, only one patient has been reported previously who was operated on at diverse spinal levels (Case 4 of Hancock and Phillips). All three of these patients (Hancock's patient and our Cases 7 and 8) exhibited significant neurological deficit at the time of operation, namely, paraplegia. One of our two patients (Case 8) had complete relief of symptoms for 10 years after initial operation, while the ensuing three operations failed to achieve any significant or lasting improvement. Extensive and multiple laminectomies did not result in any significant palliation either in our Case 7 or in Hancock's case. The fact that one of our two patients with diffuse stenosis required multiple operations within a short time indicates that the extent of spinal stenosis was not fully appreciated initially, resulting in inadequate decompression and residual compression of the neural elements. This may reflect the difficulty encountered in determining the extent of spinal stenosis radiographically, since suboptimal myelography is common in achondroplasia.

Anomalies of Intervertebral Discs

Intervertebral discs in achondroplastic patients are congenitally hyperplastic and have a tendency to bulge laterally and posteriorly. Encroachment on the spinal canal by intervertebral disc prolapse or herniation is a recognized etiological factor in paraplegia. Multiple protruding discs are common in the adult achondroplastic dwarf, and usually produce few symptoms. The presence of a prolapsed disc, coupled with a congenitally stenotic canal that has a characteristic progressive caudal stenosis, predisposes to compression of the neural elements, usually the cauda equina. Intervertebral disc degeneration is most common in the lumbar spine in the general population. However, achondroplastic patients have a tendency to disc degeneration at a distinctly higher level than in normal spines; this fact is seen in our Case 3, and in the two patients with L2–3 disc prolapse reported by Schreiber and Rosenthal. The etiology of high lumbar disc degeneration is speculative.

Nuclear prolapse from an intervertebral disc into the spinal canal in achondroplasia has been reported only five times. It is often associated with trauma or sudden stress to the back, and presents with the rapid onset of back pain and neurological deficit, occasionally with flaccid paraplegia. Our experience with Case 3 was similar to that of others. While there was no particular episode of trauma or acute strain, our patient had recently completed a full-term pregnancy with resultant increase in thoracolumbar kyphosis, and osteophyte formation. Decreased height of the intervertebral disc spaces may indicate disc degeneration with the potential for herniation. Detailed analysis of conditions within the spinal canal requires contrast myelography. Lumbar puncture, the usual route of entry into the spinal subarachnoid space, may be difficult, if not impossible, to perform in the presence of spinal stenosis. The difficulties with lumbar puncture may be compounded by the presence of a partial or complete subarachnoid block. Lumbar puncture and, therefore, lumbar myelography, is often a difficult enterprise in achondroplastic dwarfs. Indeed, the literature is replete with instances in which lumbar puncture and myelography were unsuccessful. The technique of lateral high cervical puncture for cervical myelography obviates the necessity for lumbar puncture. While cervical myelography has been used successfully in achondroplasia, even this method may be unsuccessful in certain cases where cervical arachnoiditis or basilar impression is present.

Myelography presents a distinctive picture in achondroplasia with filling defects in the contrast column at multiple levels. Often the subarachnoid space is constricted to such a degree that the contrast medium cannot flow freely. Injection of contrast
material into the subarachnoid space may be misinterpreted as a subdural or epidural injection based on the lack of characteristic flow; in this respect, even successful myelography may be an unsatisfactory means of diagnosis. Once introduced, contrast material tends to puddle in the shallow "dish" formed by the concave posterior surface of the mushroomed vertebral bodies in the prone position, further confounding diagnostic efforts.

Transverse scanning of the spine by CT has been reported, and has proven to be an effective means of diagnosing spinal stenosis. In achondroplasia and other forms of spinal stenosis, CT scanning furnishes considerable information regarding the cross-sectional anatomy of the spine at any chosen level without being invasive; however, it has the disadvantage of its inability to identify soft-tissue defects in the spine, such as a prolapsed disc, unless contrast enhancement is employed. Experimental techniques in CT scanning that allow the identification of intervertebral discs without resort to contrast use are promising; however, they are not generally available for clinical use as yet.

Three of our patients with spinal stenosis (Cases 4–6) were accurately diagnosed by CT scanning of the lumbar spine (Fig. 4). In two patients (Cases 4 and 5), the CT scan was the sole radiographic examination except plain x-ray films of the spine, and, used in lieu of myelography, accurately directed decompression laminectomy. The third patient (Case 6) underwent myelography, which confirmed the findings of the CT scan before operation. The exact extent of spinal stenosis is often difficult to estimate from radiographic examination, when incomplete or complete subarachnoid block and arachnoiditis result in suboptimal myelography. In our experience, CT scanning of the spine can accurately diagnose spinal stenosis in achondroplastic dwarfs, and greater use of this technique seems warranted, particularly in those patients in whom myelography alone is technically unsatisfactory.

Conclusions

Neurological deficits secondary to spinal stenosis represent a distinct threat to achondroplastic individuals. Of the 41 patients with achondroplasia admitted to the UCLA affiliated hospitals, 17 (41%) were admitted as a result of spinal neurological complications.

The spinal stenotic syndromes in achondroplasia may be divided into three categories: thoracolumbar, foramen magnum and upper cervical, and generalized. While the bone changes responsible for spinal stenosis are present throughout the spinal axis, the most common site of myeloradiculopathy is the lumbar spine or the thoracolumbar junction. The majority of spinal stenosis occurs in adult achondroplastic dwarfs in

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**Fig. 4.** Computerized tomography scans of the lumbar spine. *Left:* Scan at the L-1 level. Although pedicles are short, the laminar thickness and spinal canal capacity are nearly normal. *Right:* Scan at the L-5 level. Pedicles are short and thickened. Laminae are massively thickened, and the cross-sectional area of the spinal canal is markedly reduced.
spinal complications of achondroplasia

their fourth or fifth decades. Children and young adults, however, appear to account for a higher percentage of patients coming to operation than was previously suspected. Neonates and infants are susceptible to cervicomedullary compression from foramen magnum insufficiency, which may manifest itself by slowed motor development during the first year of life.

In achondroplastic dwarfs, usually those in Groups I and II, who demonstrate the insidious onset of neurological deficit over many months or years, surgical decompression of the affected neural structures can often arrest progressive neurological deficit, relieve radicular pain, and, in some instances, completely ameliorate neurological deficits. The prognosis for the patients who comprise Group III appears poor, although earlier recognition and aggressive early treatment may improve their outlook.

A thorough physical and neurological examination, as well as a detailed radiographic study, must be employed to identify correctly the level and extent of neural compromise. This will allow the surgeon to properly tailor a decompressive operation to each patient's myeloradiculopathic syndrome, with the realization that spinal stenosis may occasionally be diffuse, requiring extensive laminectomy at multiple levels. Myelography may be difficult and unreliable as a diagnostic adjunct in achondroplastic spinal stenosis, whereas CT scanning may provide a useful, non-invasive analysis of spinal stenosis in this clinical setting.

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References


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