Dwarfism secondary to achondroplasia has been recognized for centuries and pictured in historical drawings and documents. The name, according to Walter Dandy, was given to this disease after the studies of Parrot which culminated in a publication in 1878 describing the adult condition now known as "achondroplasia."

Over the centuries achondroplasia has been known to afflict these people in a distinctive manner, giving a typically enlarged head with prominent forehead, a kyphotic lumbar spine with an increased lumbar lordosis, large buttocks, and extremely short arms and legs. Achondroplastic dwarfism has been considered to be consistent with a normal life from a neurological point of view, and, except for the small pelvic outlet of achondroplastic women who bore children, the related manifestations of the disease have been poorly defined. Even today, potential neurological deficits are often not recognized early enough to forestall the irreversible changes that can be prevented by proper surgical treatment.

The cause of achondroplasia is not known; it is inheritable and is a disease involving disturbance of endochondral ossification. It occurs in dogs, notably dachshunds and bassett hounds, as well as in human beings. The disorder of endochondral bone formation affects all parts of the body, including the base of the skull. The grotesque appearance of some of these patients with hydrocephalus as described by Walter Dandy is well remembered. The short arms and legs found in these patients are the most characteristic features of the disease. The facial changes brought about by the endochondral ossification are so characteristic as to make all people with the disease appear to be related.

The disease, however, does affect the spine, though this was little recognized until the work of Donath and Vogl in 1925. On some occasions, there is stenosis of the cervical spine which results in devastating constriction of the spinal cord.

In a few instances, such as those reported by Dandy and by Dennis, et al., the head may be enlarged with hydrocephalus, which in itself will demand attention. It is interesting that Dennis and his associates found in their five cases, and in eight additional cases from the literature, that the brain's weight in all but one of 13 patients was increased over what might be expected. This has also been true in postmortem examinations of patients in whom a diagnosis has been made of premature closure of the cranial sutures. Whether this further contributes to the neurological deficit because of the constriction in achondroplasia itself is not known, but it raises an intriguing area of speculation.

The subject to be primarily considered in this paper, however, is the small lumbar spinal canal when it occurs in achondroplasia. Since the lower dorsal region is usually involved with the lumbar spine, they will be dealt with as a common entity.

Donath and Vogl by their detailed clinical studies, autopsy studies including dissection of the vertebral bodies and discs, and examination of achondroplastic fetuses, demonstrated that abnormalities of the spine are a regular occurrence in achondroplasia; changes are produced in the individual vertebra as well as in the vertebral column as a whole.

The decrease in height of the individual...
vertebral bodies accounts for the relative shortness of the whole spine. The pedicles are extremely short, particularly in the dorso-lumbar region, but to some extent also in the cervical region; this is the result of premature synostosis of the centers of ossification of the bodies with those of the laminae which are almost in contact with the dorsal part of the vertebral bodies. There is a resultant narrowing of the entire spinal canal in the anteroposterior dimension. The spinal cord, cauda equina, and conus medullaris are normal in size and are therefore subject to constriction.

As Caffey pointed out, in the spine and remainder of the skeleton in achondroplasia all centers of ossification and cartilaginous centers are too small, and there is an excess of cartilaginous centers. There is an absolute and relative increase in the cartilage of the young achondroplastic spine. This is evident even in a 1-year-old child in whom the intervertebral disc is usually as thick as the foreshortened lumbar vertebral body, whereas in a normal child at this age the disc is only one-third the size of the vertebral body. The transverse deformities of the lumbar vertebral bodies are usually overlooked and were not thoroughly appreciated until the work of Caffey. There is a progressive terminal lumbar stenosis which is exactly opposite to that seen in most lumbar spines. In a normal person, the fifth lumbar vertebra is the widest by far, but in the achondroplastic lumbar spine the fifth lumbar body is the narrowest. The spinal canal is therefore not only narrowed in the anteroposterior direction but also in the transverse direction, so that the fifth lumbar vertebra is narrower than the first, and the spinal canal tapers progressively downward.

The shape of the lumbar and of the dorso-lumbar segment of the spine becomes even more important with advancing age. Some degree of dorsolumbar kyphosis is a part of the achondroplastic process, but the occasional gibbous formation is a complication which occurs in postnatal life. Donath and Vogl classified the shape of the dorsi-lumbar portion of the spine in achondroplasia into four types: 1) flat back, 2) moderately kyphotic, 3) extremely kyphotic, and 4) sharply angular.

The gibbous formation in the fourth type is usually caused by the presence of one and sometimes two wedge-shaped vertebral bodies between the twelfth thoracic and second lumbar vertebra.

As Epstein and Malis pointed out, the decrease in the height of the vertebral body results in shortening of the spine, while shortness of the pedicles leads to constriction of the canal, greatest in the anteroposterior diameter. The thick pedicles are deformed by the premature synostosis of the centers of ossification of the body with those of the lamina. The canal of the adult achondroplastic dwarf is narrowed by marginal exostoses and sometimes further by protrusion of the intervertebral discs.

Duvoisin and Yahr further emphasized this and showed that, in addition to the thickening of the cartilaginous plates at the edge of the vertebral bodies and the narrowed spinal canal, the laminae are often thickened, even further limiting the spinal canal.

It is rare for neurological changes in the thoracolumbar region to occur in childhood or early adulthood. Since the spinal cord is of normal size as are all the neural elements in achondroplasia, these elements fit snugly into the small spinal canal and any progressive encroachment with age on the lumen of the spinal canal has a major mechanical effect on the spinal cord and the intraspinal nerves.

The dorsi-lumbar segment of the achondroplastic spine is an unstable portion, often with a pronounced dorsal kyphosis, and there are frequently large osteoarthritic spurs as well as spur formations of the articular processes. These changes further increase the constriction of the lumen of the spinal canal, and in some cases actual free rupture of the intervertebral discs has occurred.

The numerous changes found in human achondroplasia are dramatically demonstrated in studies of typically achondroplastic dogs, primarily the dachshund, the bassett hound, the French bulldog, and the Pekingeses. Dogs have shown a high incidence of disc degeneration and in many instances free protrusion and extrusion of the discs into the small spinal canal. The narrowing of the spinal canal in dogs is just as evident as in human achondroplasia, and paraplegia with remissions and exacerbations