Spinal Stenosis in Achondroplasia

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Achondroplasia is the commonest form of short-limbed dwarfism. It is transmitted as an autosomal dominant with a high mutation rate, with the result that the majority of affected persons are born to normal parents with no previous family history. One in 2 of the children born to an achondroplastic parent will be affected. Its incidence has been estimated to vary between 17 and 29 per million population; i.e. in the United Kingdom there are between 850 and 1500 affected individuals. The etiology is unknown; raised paternal age has been shown to be a significant factor.

It produces a typical clinical picture which varies only slightly from patient to patient. The limbs are very much shortened in contrast to the spine, which has an essentially normal height. Because of this it has been assumed for years that the spine is spared in this disorder. However, in 1925 Donath & Vogl pointed out that there are a number of very important structural changes in the vertebral column. In fact these abnormalities in the spine account for the most serious complication in achondroplasia, namely spinal cord and nerve root compression. Donath & Vogl collected 9 reported cases of this complication and added a tenth. Since their report, about 30 cases have been recorded.

The primary defect is a failure in growth occurring in the proliferative zone of the growth plate. This results in an initially small cartilaginous anlage and a subsequently short thick long bone. A vertebra is preformed in cartilage and ossifies from three primary centres, one for the body and one for each half of the posterior arch. Impaired longitudinal growth at the latter centres causes a shortening of the pedicles and a reduction of the interpedicle distance. Donath & Vogl (1925) attributed these abnormalities to premature synostosis, but for this statement there is little evidence; simple impaired growth would account for the shortening (L O Langer 1969, personal communication).

Another important area which is affected by the defect in cartilage proliferation is the base of the skull, which is almost entirely preformed in cartilage. Reduction in the size of the foramen magnum and associated abnormalities of the atlas and axis may occur.

At birth, the achondroplastic spine has the normal 'C' curve of the flexed embryo. As the child begins to sit up, the secondary curve in the cervical region develops. The large size of the head, together with the general hypotonia, results in a postural kyphosis which appears at the lower thoracic or the upper lumbar region. This is usually temporary and in most patients disappears once walking occurs. However, in some patients this kyphosis persists and may progress to become a structural abnormality with wedging of the apical vertebræ. Mørch (1941), in a review of 81 patients, reported a persistence of this kyphosis into adulthood in one-third of his series.

Associated with the attainment of the upright posture is the development of the lumbar lordosis. This is excessively developed in the achondroplastic and increases as adulthood is reached. The shape of the vertebral body also shows a progressive change with age. This is particularly evident at the posterior border of the body. Within months of birth there is a progressive excavation of the posterior surface of the vertebral body, resulting in the classical scalloping seen in the adult. Although recorded by many authors, no explanation for this appearance has been previously suggested. The progressive excavation of the posterior surface of the vertebral body is due to the pressure created by the normal spinal contents in the presence of the short pedicles; it is the only available way of enlarging the canal.

Spinal stenosis may be defined as a disproportion between the normal spinal contents and the bony spinal canal. Vogl & Osborne (1947) stressed the presence in achondroplasia of generalized stenosis, due to the short pedicles and the reduced interpedicle distance. This may be further compromised by a small foramen magnum, a proximally displaced atlas or axis, a structural kyphosis, commonly in the thoracolumbar region, prolapse of an intervertebral disc, hypertrophic osteoarthritis of the posterior joints and an increased lumbar lordosis. Symptoms of spinal cord or nerve root compression rarely present before the age of 15, and at this age only when a significant kyphosis is present. More commonly they come on towards the end of the third and in the fourth decade.

Clinical Material

Seventy-one patients were reviewed, 40 male and 31 female; their ages ranged from 3 months to 68 years. Spinal complications were common, being present in 33 (46%). The patients were divided into three groups, according to age.

Group 1, aged under 15 (51 patients): Of these 13 had a spinal deformity. Two had idiopathic scoliosis which corrected spontaneously before the age of 3 (and was probably unrelated to the achondroplasia); 11 (20%) had thoracolumbar kyphosis, which corrected spontaneously in 4 but increased in 7, 2 of whom developed cord com-

pression. Of the 7 patients in whom kyphosis increased, no details were available about 2, in 3 the condition was controlled by means of a brace or spinal fusion, and 2 deteriorated to the point where cord compression was present and decompression was carried out, with some improvement in both cases.

Group 2, aged 15–29 (7 patients): Of these 3 had no symptoms, 1 had backache with no neurological signs, 1 had a disc prolapse which responded to bed rest, 1 had kyphosis with paraplegia which responded to posterior decompression, and 1 had a paraparesis due to spinal stenosis which responded to bed rest.

Group 3, aged 30 and over (13 patients): One patient had backache, 2 had backache with CNS signs, 8 had a paraparesis and 2 had a quadriparesis. Of the 8 patients with a paraparesis, 2 responded to rest and back support, 1 made an excellent recovery following laminectomy and 1 elderly patient with a complete spinal block was unfit for surgery; in 4 no details of treatment were available. Of the 2 patients with quadriparesis, 1 improved considerably and was able to walk with aids following posterior cervical fusion; the other made an initially good response following cervical and lumbar laminectomy for spinal stenosis, but deteriorated later.

Four clinical syndromes may be observed:
(1) Single and multiple disc lesions, usually in the lumbar region and presenting with a typical history and findings.
(2) Generalized spinal stenosis, often beginning in the lumbar region with a history of intermittent claudication. Later the upper limbs become involved and quadriparesis develops.
(3) Severe thoracolumbar kyphosis with spinal block; clinically, progressive kyphosis with associated cord and root signs.
(4) Foramen magnum insufficiency: compression of the medulla and upper cervical spine due to a small foramen magnum and a proximally placed atlas and axis, resulting in quadriparesis.

Management
Results of surgery in these syndromes have been reported as follows.
Single and multiple disc prolapse (Schreiber & Rosenthal 1952, Spillane 1952): Three of the 4 reported cases did well following laminectomy and disc removal; the fourth was found to have multiple disc prolapses and the result was an improvement only.
Generalized spinal stenosis: Seven patients are reported (Duvoisin & Yahr 1962, Hancock & Phillips 1965). Results were variable and seemed to depend on early and adequate surgical decompression; the stenosis was often more extensive than originally thought and repeated surgical decompression was required. Very detailed preoperative assessment is necessary if the true extent and severity of the lesion is to be appreciated.
Thoracolumbar kyphosis: Six patients with a significant thoracolumbar kyphosis associated with neurological signs and having undergone surgery are reported (McCluer 1961, Duvoisin & Yahr 1962, Hancock & Phillips 1965, Alexander 1969). The youngest, a child of 5, underwent two attempts at fusion of the unstable segments. He remained well until aged 28, when he developed progressive weakness of the legs, associated with a spinal block; laminectomy resulted in little improvement. The results were generally disappointing, but one patient did well following wide decompression from T12 to L5 (Alexander 1969).
Foramen magnum insufficiency: One patient in this group failed to improve following two surgical decompressions (Hancock & Phillips 1965).

Conclusion
Three important principles emerge from a review of the surgical results.
(1) A progressive kyphosis in a child requires urgent splintage. If the kyphosis continues to progress then fusion is required.
(2) The significance of early symptons and signs in spinal stenosis must be stressed and early decompression performed, as results are very disappointing once extensive neurological damage has occurred.
(3) Finally, careful preoperative assessment of the whole spinal canal is necessary if surgical treatment is to be adequate.

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