

Computed Tomography of the Foramen Magnum: Achondroplastic Values Compared to Normal Standards

Achondroplasia, the most common of the skeletal dysplasias, is an autosomal dominant disorder whose clinical manifestations include short-limbed dwarfism, large head, shallow thoracic cage, and characteristic radiographic findings. This disorder is characterized by a decreased rate of endochondral ossification and normal membranous ossification.

The foramen magnum has long been recognized to be small in achondroplastic individuals. The small size is secondary to deficient growth of the endochondral exoccipital, supraoccipital, and basioccipital bones, which form the boundaries of the foramen magnum. Neurologic abnormalities seen in achondroplastic patients have included respiratory embarrassment, quadripareis and paraparesis, obstructive hydrocephalus, and sudden death. Compression of the upper cervical spinal cord and caudal medulla due to the small foramen magnum has been seen at autopsy in several cases. Surgical decompression by suboccipital craniectomy and cervical laminectomy has been suggested for patients with evidence of neurological involvement.

In this report, the extent of foramen magnum stenosis in achondroplasia was quantified by measurement of the maximal transverse and saggittal lengths of the foramen magnum by computed tomography (CT) scan. These were compared against foramen magnum measurements for persons of normal stature.

CT scans of the foramen magnum were performed by scanning at 0° horizontally from the top of the hard palate through the foramen magnum to the occiput. Maximum transverse and saggittal foramen magnum length was measured on the axial bone window scan with a ruler. Measurements were obtained from people of normal stature and varying ages: the transverse dimension was measured in 164 cases and the saggittal dimension in 144. Mean normal values ± 1 SD were calculated at monthly intervals from birth

through 2 years of age and at two-year increments thereafter. It was found that the mean foramen magnum size did not change appreciably after 15 years of age.

These values were then compared with foramen magnum measurements from 63 patients with achondroplasia. Among this group, 41 patients had no neurological findings. Twenty-two had evidence of neurological dysfunction suggestive of foramen magnum compression based on history, physical exam, short latency somatosensory potentials, and/or polysomnography.

From the data, one can conclude that the normal foramen magnum grows rapidly in both dimensions from birth to 1 year of age and then continues at a greatly diminished rate until approximately 15 years of age. The foramen magnum in achondroplastic individuals was significantly smaller than that of normal people at all ages. Achondroplastics without neurological dysfunction had measurements within ± 5 SD of the normal mean for the transverse and ± 4 SD for the saggittal dimensions. Patients with neurological symptoms had significantly smaller measurements. Included in this latter group were seven patients with extremely small foramen magnum size and obstructive hydrocephalus. Significantly, foramen magnum size was not shown to correlate with head circumference in this or any other group studied.

The authors concluded that stenosis of the foramen magnum may be more widespread in achondroplasia than had been previously appreciated. Because significant morbidity and potential mortality are associated with this stenosis, CT scans may identify individuals at high risk for these complications. However, the efficacy and safety of surgical decompression has not yet been clearly documented. The authors recommend that CT scan of the foramen magnum be considered part of the comprehensive care of individuals with achondroplasia.

Hecht JT, Nelson FW, Butler IJ, et al: *Am J Med Genet* 1985;20:355.

Editor's comment—*The recent recognition of late infantile mortality as a significant complication of*

achondroplasia has made careful neurological surveillance of achondroplastic children imperative. This report documents the differences in foramen magnum dimensions between normal and achondroplastic children, and especially between achondroplastic children with and without neurological complications. Newer diagnostic imaging procedures, such as magnetic resonance imaging (MRI) scans, have also become useful in the evaluation of spinal cord compression due to upper cervical vertebrae or foramen magnum stenosis in achondroplasia or other skeletal dysplasias. Unlike CT scanning, MRI can easily distinguish among varying soft tissue densities and can identify kinking or impingement on the spinal cord at an early stage. Discovery of anatomic abnormalities should be followed up by neurological and neurophysiological evaluation, including somatosensory-evoked potentials. Definitive criteria for neurosurgical intervention, however, remain to be established.

Impaired Calcitonin Secretion in Patients With Williams Syndrome

The Williams syndrome (WS) is characterized by prenatal and postnatal growth retardation, microcephaly, facial dysmorphism (the so-called elfin facies), congenital heart disease (most commonly, supravalvular aortic stenosis), and mental deficiency. In a number of patients, this condition has been associated with neonatal hypercalcemia. This latter finding led to the initial description in 1952 of WS as "idiopathic hypercalcemia of infancy." In older children, metastatic calcium deposits have been found in the kidney, and osteosclerosis has been seen on x-ray in the skull and the metaphyses of the long bones.

Because the etiology of the hypercalcemia seen in WS has been debated for a number of years, the authors examined several aspects of calcium and vitamin D metabolism in five children with WS and seven age-matched controls. At the