Neurosurgical Management of Congenital Malformations and Inherited Disease of the Spine

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KEYWORDS

- Achondroplasia • Goldenhar syndrome
- Lipomyelomeningocele • Morquio syndrome
- Myelomeningocele • Osteogenesis imperfecta
- Spina bifida • Spondyloepiphyseal dysplasia

Congenital malformations encompass a diverse group of disorders present at birth, as result of genetic abnormalities, infection, errors of morphogenesis, or abnormalities in the intrauterine environment. Congenital disorders affecting the brain and spinal cord are now often diagnosed before delivery with the use of prenatal ultrasonography and maternal serum screening. Over the past several decades there have been major advances in the understanding and management of these conditions. This article focuses on the most common spinal congenital malformations, limiting the discussion to the neurosurgically relevant aspects of myelomeningocele, lipomyelomeningocele, and skeletal dysplasias, including achondroplasia, Goldenhar syndrome, Morquio syndrome, spondyloepiphyseal dysplasia (SED), osteogenesis imperfecta (OI), and Larsen syndrome.

Skeletal dysplasias are a heterogeneous group of more than 200 disorders in which there is abnormal formation, growth, or remodeling of cartilage and bone. This group of disorders affects 1 in 5000 live births, differing widely in natural history, inheritance, and etiology. Skeletal dysplasias may be classified as either osteochondral dysplasia (involving the whole skeleton) or dysostosis (involving a single group of bones). Craniocervical junction abnormalities, atlantoaxial subluxation, and kyphoscoliotic deformities are common spinal problems found in skeletal dysplasias. This discussion focuses on the key skeletal dysplasias and their neurosurgical implications in children.

ACHONDROPLASIA

Achondroplasia is the most common form of dwarfism and is the most common heritable skeletal dysplasia, characterized by a disproportionate shortening of the proximal limbs relative to the

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trunk. This is sometimes referred to as rhizomelic dwarfism, describing the small forearms and thighs in relation to the entire limb and trunk. It occurs in 1 of every 28,000 live births.[1–3] Recognizable at birth, the characteristic morphology includes shortened limbs and long bones, macrocephaly, frontal bossing, genu varum abnormalities, and a low-set nasal bridge.[4] Achondroplasia follows an autosomal dominant inheritance pattern, although 80% of cases arise as a result of spontaneous mutations in the fibroblast growth factor receptor 3 (*FGFR3*) gene located on chromosome 4.[1,2,5] This gene mutation results in a decrease in the rate of endochondral bone formation with normal rates of membranous bone formation, calcification, and remodeling.[4,5]

Almost half of all children with achondroplasia have neurologic manifestations of their disease.[2] The neurologic manifestations of achondroplasia include ventriculomegaly, compressive spinal syndromes, and developmental delay.[6] Other than ventriculomegaly, foramen magnum stenosis resulting in cervicomedullary compression is the most frequent cause of neurosurgical consultation in infants. In older children and adults, multisegment spinal stenosis involving the subaxial cervical or thoracolumbar spine may also be present. Foramen magnum stenosis results from abnormal endochondral bone growth and fusion of posterior basal synchondroses.[3,7] The bones of the skull base, as well as bones of the neural arches, normally enlarge by endochondral ossification.[8,9] Because of defective endochondral bone formation at the cranial base and craniocervical junction, infants with achondroplasia may have a small foramen magnum, a short basicranium and clivus, a shallow posterior fossa with a horizontally oriented inferior occiput, an abnormal odontoid process, stenotic jugular foramina, and a narrow upper cervical canal.[1,3,8] Furthermore, premature fusion and abnormal development of the 2 posterior synchondroses contribute to thickening of the rim of the foramen magnum, which may project into the brainstem causing compression and severe angulations of the medulla and rostral cervical spinal cord.[1,8] The odontoid often projects superiorly and posteriorly into the small foramen magnum causing the anterior medulla to drape over the odontoid.[8,9] These changes lead to damage of the corticospinal tract and chronic ischemia to the medulla.[8]

Although foramen magnum stenosis is found in as many as 70% of achondroplasia patients, only between 10% and 35% of patients exhibit symptoms of cervicomedullary compression.[10,11] Cervicomedullary compression secondary to foramen magnum stenosis can present with dysfunction of the lower brainstem, high cervical spinal cord, and associated nerve roots in children with achondroplasia. Symptoms of cervicomedullary compression may include poor head control, excessive hypotonia, apnea, feeding difficulties, developmental delay, hydrocephalus, myelopathy, respiratory disorders, and sudden death due to respiratory arrest.[12] Less lethal respiratory disturbances in children with achondroplasia include central and obstructive sleep apnea.[13] Central sleep apnea in these children is thought to result from damage to the ventral medullary respiratory control centers from foramen magnum stenosis. Foramen magnum stenosis may also compress lower motor neurons leading to paralysis of the diaphragm and accessory muscles of respiration.[1] Apnea due to foramen magnum stenosis may improve dramatically after surgical decompression.[7,13–15] Hydrocephalus in children with achondroplasia is theorized to result from a combination of crowding of the foramen magnum, resulting in obstruction of cerebrospinal fluid (CSF) outflow, and jugular foramen stenosis, resulting in elevated venous sinus pressures.[9,13] In many instances, the ventriculomegaly found in children with achondroplasia arrests without treatment. Shunt placement can be considered for those who have failed conservative treatment.[1,13,14] Even without hydrocephalus, macrocephaly is extremely common in achondroplasia and relative head size should be assessed compared with disease-specific normative data.

The decision to recommend surgical decompression of foramen magnum and cervical stenosis in achondroplasia patients should be based on both clinical signs and symptoms and imaging data. The clinical signs and symptoms of most concern include lower cranial nerve palsies, apnea, hyperreflexia, clonus, and weakness. Concerning findings on magnetic resonance (MR) imaging include intramedullary T2-weighted changes, absence of CSF signal at the foramen magnum, and the presence of a syrinx. One prospective study of children with achondroplasia concluded that the signs and symptoms that best predicted the need for surgical decompression included central hypopnea, foramen magnum measurements below the mean for children with achondroplasia, lower-extremity hyperreflexia, and clonus.[10] Foramen magnum decompression via a suboccipital craniectomy with removal of the posterior arch of the atlas is performed in cases of proved symptomatic cervicomedullary compression. Even in apparently asymptomatic children, the presence of significant stenosis and T2 signal change in the spinal cord is strongly suggestive of the need for decompression or