Best Practice Guidelines for Management of Spinal Disorders in Skeletal Dysplasia

Disorders of the spine are a significant cause of morbidity and mortality in skeletal dysplasia.[1,2,3] Spine disorders may present early in life and are often progressive and severe. Spinal cord compression, with resultant paralysis or death, is the greatest concern. Treatment does not always follow established norms for spinal disease. Spinal disorders include kyphosis, scoliosis, central stenosis, vertebral instability, and hyperlordosis, often resulting in neurologic decompensation. Careful neurological examination should be regularly performed to assess for the presence of myelopathy. Symptoms or signs of myelopathy in an individual with skeletal dysplasia require further evaluation, as catastrophic injury can occur.[4] Once the level of myelopathy is identified, appropriate steps to manage the spinal cord and canal structure can be determined.

Common vertebral shape anomalies in the context of skeletal dysplasia include platyspondyly, anterior vertebral body beaking, coronal clefting, narrowing of the interpediculate distance in lumbar vertebrae, and posterior scalloping of the vertebral body. Anterior vertebral body beaking is typical of pseudoachondroplasia and the mucopolysaccharidoses (MPS). When beaking is present, MPS should be considered and diagnosis pursued to rule out these treatable disorders.

Anyone with a skeletal dysplasia that has known hypoplasia of the odontoid, axis, or other cervical vertebrae is at risk for instability that could predispose to spinal cord injury. As soon as a young child can cooperate to perform flexion-extension radiographs of the cervical spine (typically age 2–3 years), imaging should be done to define cervical spine stability. Flexion-extension MRI of the cervical spine under sedation or anesthesia is helpful in assessing cervical instability.[5] Three-dimensional imaging and angiography with CT and/or MRI are strongly recommended to accurately assess anatomy and plan for surgery.

Non-surgical management may be appropriate for some spinal complications, but close observation is needed to assess if and when surgery is indicated. These measures include physical therapy, weight reduction, and the use of appropriate non-steroidal anti-inflammatory analgesia. Since the neurological complications of bowel dysfunction, urinary incontinence, and spasticity may not recover following surgery, the severity should not be permitted to progress to the point that function cannot be restored. Bracing with thoraco-lumbar-sacral orthoses (TLSOs) has been used in the treatment of thoracolumbar kyphosis, particularly in patients with achondroplasia.[6] To minimize the likelihood of vertebral body wedging, anticipatory guidance regarding posture in the care of these infants is recommended in early infancy.[7] For scoliosis,

young patients with skeletal dysplasia and flexible spinal deformity respond well to bracing and serial casting.

Surgical intervention for cervical instability and spinal stenosis should be reserved for demonstrable pathology or symptoms rather than performed prophylactically. Surgical intervention for thoracolumbar kyphosis should be reserved for deformities that are progressive or symptomatic. In progressive scoliosis, treatment paradigms are similar to other conditions with scoliosis. Growth-friendly surgery is appropriate in patients with skeletal dysplasia in accordance with principles of early-onset scoliosis management.[8]

There are reports of spinal cord infarct after prolonged neurosurgical or orthopedic procedures, in anatomic areas separate from the primary site of operation.[9,10,11,12] The area of greatest risk for injury appears to be at the upper thoracic level. Neuromonitoring should be performed for all surgeries exceeding one hour, including all non-spine surgeries.

Table 1: Skeletal Dysplasia with Significant Spinal Manifestations*

Group/Name of Disorder	Inheritance	Gene	OMIM Number	ORPHANET Code	Typical Spinal Manifestations†
Achondroplasia	AD	FGFR3	100800	18060	2,3,5
Hypochondroplasia	AD	FGFR3	146000	146000	5
Spondyloepiphyseal dysplasia congenita (SEDC)	AD, AR	COL2A1	183900	604864 616583	1,3,4
Kniest dysplasia	AD	COL2A1	156550	485	1,3,4
Diastrophic dysplasia (DTD)	AR	SLC26A2	222600	628	1,3,4
Atelosteogenesis type 3 (AO3)	AD	FLNB	108721	56305	3,4
Larsen syndrome (dominant)	AD	FLNB	150250	503	3,4
Metatropic dysplasia	AD	TRPV4	156530	2635	1,2,3,4,5
Pseudoachondroplasia (PSACH)	AD	COMP	177170	750	1,4
Campomelic dysplasia (CD)	AD	SOX9	114290	140	2,3,4
CDP, X-linked dominant, Conradi–Hünermann type (CDPX2)	XL	EBP	302960	35173	1,3,4
Osteogenesis imperfecta, progressively deforming type (OI type 3)	AD	COL1A1 COL1A2	259420	216812	4
Mucopolysaccharidosis type 1H	AR	IDUA	607014	579	2,3
Mucopolysaccharidosis type 4A	AR	GALNS	253000	309297	1,3,4
Mucopolysaccharidosis type 6	AR	ARSB	253200	583	1,2,3

* Adapted from Mortier et al. 2019

† 1: cervical instability, 2: cervical stenosis, 3: cervical/thoracic/thoracolumbar kyphosis, 4: scoliosis, 5: lumbar stenosis

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