Coffin Lowry syndrome

Coffin-Lowry syndrome (CLS) is a rare genetic disorder inherited in an X-linked dominant pattern. Common manifestations include intellectual disability, growth retardation, dysmorphic facial features, and variable skeletal anomalies. Here we report a patient who first presented with episodes of apparent life-threatening events (ALTE) found to be caused by hydrocephalus and brainstem compression at the foramen magnum. Together with his small size, short limbs and fingers, and facial appearance, the narrowing of the foramen magnum lead to the initial clinical misdiagnosis of hypochondroplasia. Subsequent evaluation and testing lead to the correct diagnosis of CLS. This case demonstrates the variability in presentation of CLS, and that skeletal findings may be misleading in infancy [1].

Morino et al., report 8 years of follow-up after decompression to treat cervical myelopathy in a patient with Coffin-Lowry syndrome (CLS). CLS is

In this patient, the spinal cord was compressed by calcium pyrophosphate crystal deposition in the cervical yellow ligament (YL). To date, only 1 report has described clinical features after surgery for calcified cervical YL in CLS.

A 15-year-old male with tetraplegia secondary to compression of the cervical spinal cord induced by a hypoplastic posterior arch of C1 and calcification of the YL from C2 to C7 was treated surgically with laminectomy from C1 to C7. The patient's history, clinical examination, imaging findings, and treatment are reported. The patient was incapable of speech because of mental retardation, so he could not describe his symptoms. Gait disturbance worsened over the 2 months before admission to our hospital. At admission, the patient could not move his extremities, and tendon reflexes of the upper and lower extremities were significantly increased. Computed tomography of the cervical spine showed YL calcification from C2 to C7. Magnetic resonance imaging showed consecutive compression of the cervical spinal cord. We diagnosed quadriplegia secondary to cervical cord damage and performed emergency surgery.

During C1-C7 laminectomy, YL calcification in C2-C7 was observed. The calcification was confirmed as calcium pyrophosphate by crystal analysis. Quadriplegia gradually resolved, and almost disappeared by 2 weeks after the operation. Cervical hyperlordosis was observed in radiographs starting from 1 month after the operation, but it has not progressed and is not associated with any symptoms.

The efficacy of decompression continued, and no postoperative complications have occurred during at least 8 years of follow-up [2].

1)
Upadia J, Oakes J, Hamm A, Hurst AC, Robin NH. Foramen magnum compression in Coffin-Lowry syndrome: A case report. Am J Med Genet A. 2017 Feb 12. doi: 10.1002/ajmg.a.38095. [Epub ahead of print] PubMed PMID: 28190284.
2)
Morino T, Ogata T, Horiuchi H, Yamaoka S, Fukuda M, Miura H. Eight years of follow-up after laminectomy of calcium pyrophosphate crystal deposition in the cervical yellow ligament of patient with Coffin-Lowry syndrome: A case report. Medicine (Baltimore). 2016 Aug;95(31):e4468. doi: 10.1097/MD.0000000000004468. PubMed PMID: 27495083; PubMed Central PMCID: PMC4979837.