OP39

Craniovertebral junction pathological features and their management in the mucopolysaccharidoses

Erik Pietro Sganzerla[1], Andrea Trezza[1], Alessandro Versace[1], Grimaldi Marco[2], Rossella Parini[3], Carlo Giorgio Giussani[1]

[1] *Clinica Neurochirurgica, Università di Milano Bicocca, Ospedale San Gerardo, Monza, Italy*

[2] *Neuroradiologia, IRCCS Istituto Clinico Humanitas, Rozzano (MI), Italy*

[3] *Centro "Fondazione Mariani" per le Malattie Metaboliche, U.O.S. Malattie Metaboliche Rare - Clinica Pediatrica, Azienda Ospedaliera San Gerardo, Monza, Italy*

Introduction: The mucopolysaccharidoses (MPS) are multisystemic inherited metabolic diseases caused by the deficiency of the enzymes involved in the degradation of glycosaminoglicans. Despite new medical therapies positively affecting the natural history of MPS, the presence of spinal abnormalities and deposition of glycosaminoglicans in soft tissues remains nearly unaltered with possible slowly progressive myelopathy or acute post-traumatic tetraplegiaWe present the neuroimaging pathological findings in a consecutive series of 42 MPS patients.

Methods: The population consists of 12 MPS I, 15 MPS II, 2 MPS III, 9 MPS IV and 4 MPS VI. MRI studies have been scored considering: dens dysplasia, periodontoid tissue thickening, spinal stenosis, myelopathy and instability.

Results: CVJ abnormalities were frequent with a reduced diameter of the spinal canal at the CVJ in 17/42 children. The most severe spinal canal stenosis and cord compression were observed in MPS IV and MPS VI. Reduction > 50% of the cervical canal diameter was observed in 5/42 patients. MRI signs of myelopathy were present in 3/42 cases, all affected by MPS IV. Dens hypoplasia was overall present in 33/42 cases and was a constant feature in MPS IV and VI. Slight modifications of canal diameter during dynamic MRI studies were observed in 9/32 cases, confirming how clear-cut radiological signs of instability are seldom documented in MPS patients. Increased canal stenosis during dynamic MRI studies was evidenced in 6/9 MPS IV patients.

Conclusion: CVJ abnormalities have to be recognized early in MPS as unrecognized cord compression at the CVJ can result in progressive or sudden traumatic cord damage. The evidence of acute or progressive myelopathy associated with radiological spinal cord compression at the CVJ is an indication to surgery.Preventive surgery should be considered in asymptomatic patients, especially in MPS IV, when MRI studies evidence cord compression.