P-057 - “GROWING PAINS”: THE CHALLENGES OF GETTING OLDER WITH MUCOPOLYSACCHARIDOSES

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INTRODUCTION: The mucopolysaccharidoses are caused by deficiencies in enzymes involved in the sequential degradation of glycosaminoglycans (GAGs), which are ubiquitous in connective tissues. The resulting impaired degradation of GAGs in cells and tissues leads to substrate accumulation causing progressive multi-organ dysfunction. Seven types of mucopolysaccharidosis (MPS) disorders have been described, with MPS III and MPS IV each having two or more biochemical subtypes.

OBJECTIVES: To report a cohort of adult Brazilian MPS patients and their clinical aspects.

MATERIAL AND METHODS: Clinical, biochemical and radiological features of 13 adult patients with MPS ( 6 males; 7 females) were examined. MPS disorders are multi-organ diseases that require multi-disciplinary management involving clinical geneticists, metabolic physicians, orthopedists, general surgeons, neurosurgeons, pulmonologists, cardiologists, neurologists, anesthesiologists, otolaryngologists, ophthalmologists, dentists, transplant physicians, psychiatrists, and physiotherapists. Skeletal and joint abnormalities are observed in all patients but one (an adult with MPS III), most complaints were from hip pain, and difficulties to walk longer distances (mainly patients with MPS VI A). Most patients remain unemployed, living with their relatives and without going to college. Depression and anxiety were common features, in particular in two patients with MPS VI and MPS II, both with normal IQ. All patients referred that adult clinics were not aware of MPS disorders and multidisciplinary care was not possible.

CONCLUSION: The management of MPS in an adult clinical setting creates challenges due to the patients' short stature and medical complexity, the fact that most internists are unfamiliar with MPS disorders, and the limited availability of assistance for multi-disciplinary patient care (coordination of timely, patient-friendly, integrated care by different specialists) in adult clinics.