Special Issue

Mucopolysaccharidosis-Plus: From Molecular Defects to Therapeutic Strategies

Message from the Guest Editor

Mucopolysaccharidoses (MPS) are a group of lysosomal storage disorders characterized by the absence or markedly reduced activity of lysosomal enzymes involved in the degradation of glycosaminoglycans (GAGs). An MPS-like disease has recently been described and called mucopolysaccharidosis-plus syndrome (MPSPS). MPSPS represents one of the rarest diseases, with only a few dozen cases reported worldwide.

The primary pathogenic mechanism in MPS involves the accumulation of GAG(s) within lysosomes. In MPSPS, a similar accumulation occurs yet, no studies have demonstrated reduced activity of any known lysosomal enzymes. Due to the markedly shortened lifespan of affected individuals and the complex pathomechanism of the disease, no effective treatment has been developed for patients with MPSPS. Current therapeutic approaches are limited to symptomatic management. This special issue is, therefore, devoted to any kind of works on MPSPS, from basic to clinical studies, from diagnostic to translational works, from subjects related to patients' care to development of novel therapies. Both original and review manuscripts are welcome, provided they are directly or indirectly related to MPSPS.

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