Mucopolysaccharidosis

Mucopolysaccharidosis refers to a group of inherited conditions in which the body is unable to properly breakdown mucopolysaccharides (long chains of sugar molecules that are found throughout the body).

Mucopolysaccharidosis type I-H (MPS I-H) is a rare lysosomal storage disorder caused by α-L-Iduronidase deficiency. Early haematopoietic stem cell transplantation (HSCT) is the sole available therapeutic option to preserve neurocognitive functions. We report long-term follow-up (median 9 years, interquartile range 8-16.5) for 51 MPS I-H patients who underwent HSCT between 1986 and 2018 in France. 4 patients died from complications of HSCT and one from disease progression. Complete chimerism and normal α-L-Iduronidase activity were obtained in 84% and 71% of patients respectively. No difference of outcomes was observed between bone marrow and cord blood stem cell sources. All patients acquired independent walking and 91% and 78% acquired intelligible language or reading and writing. Intelligence Quotient evaluation (n = 23) showed that 69% had IQ ≥ 70 at last follow-up. 58% of patients had normal or remedial schooling and 62% of the 13 adults had good socio-professional insertion. Skeletal dysplasia as well as vision and hearing impairments progressed despite HSCT, with significant disability. These results provide a long-term assessment of HSCT efficacy in MPS I-H and could be useful in the evaluation of novel promising treatments such as gene therapy [1)].

1)
Gardin A, Castelle M, Pichard S, Cano A, Chabrol B, Piarroux J, Roubertie A, Nadjar Y, Guemann AS, Tardieu M, Lacombe D, Robert MP, Caillaud C, Froissart R, Leboeuf V, Barbier V, Bouchereau J, Schiff M, Fauroux B, Thierry B, Luscan R, James S, de Saint-Denis T, Pannier S, Gitiaux C, Vergnaud E, Boddaert N, Lascourreges C, Lemoine M, Bonnet D, Blanche S, Dalle JH, Neven B, de Lonlay P, Brassier A. Long term follow-up after haematopoietic stem cell transplantation for mucopolysaccharidosis type I-H: a retrospective study of 51 patients. Bone Marrow Transplant. 2022 Dec 9. doi: 10.1038/s41409-022-01886-1. Epub ahead of print. PMID: 36494569.