Clinical Trial of Gene Therapy for Mucopolysaccharidosis type VI - a severe lysosomal storage disorder

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MPS VI, also known as Mucopolysaccharidosis VI or Maroteaux-Lamy syndrome, belongs to a group of rare inherited diseases called lysosomal storage disorders.

Most lysosomal storage disorders are due to a genetic deficiency of an enzyme in subcellular organelles called lysosomes. When the enzyme is missing, lysosomes cannot break down a specific kind of material that accumulates and causes all of the problems seen in lysosomal storage disorders.

MPS VI is caused by a deficiency of the ARSB enzyme, which results in intra-lysosomal storage and increased urinary excretion of partially degraded glycosaminoglycans (GAGs), previously known as mucopolysaccharides. No single symptom defines MPS VI. A patient with MPS VI may develop a cluster of several symptoms affecting various body systems, with the exception of the central nervous system. Symptoms of MPS VI are not usually evident at birth, but show up later as GAGs build up. However, the rate at which symptoms appear and worsen varies widely. Some affected individuals have a rapidly advancing form of MPS VI and may start showing symptoms as early as 6 to 24 months of age. Others have a more slowly progressing form of MPS VI and may not show significant symptoms until much later. MPS VI often produces a range of recognizable changes in physical appearance, as the thickening of nose, lips, and tongue that occurs as GAGs build up in tissues. MPS VI individuals may have a large head, a protruding abdomen, and short stature.

At the moment, the recommended therapy for MPS VI is Enzyme Replacement Therapy (ERT): it consists of weekly intravenous infusions with recombinant ARSB, which last few hours.

Gene therapy has the potential to convert the liver into a factory and a “depot” for the sustained systemic release of the enzyme ARSB.