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The impact enzyme replacement therapy on growth and pubertal development in two Japanese siblings with mucopolysaccharidosis type VI

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In this case report, two siblings with MPS VI started enzyme replacement therapy (ERT) with weekly infusions of recombinant human ASB (Galsulfase) at 1 mg/kg. Sibling 1 started ERT 5.6 years of age and Sibling 2 was 6 weeks old. The disease status in these two siblings prior to and for no less than 120 months of ERT was followed up and compared. The treatment was well tolerated by both siblings. During 120 months of ERT, symptoms typical of MPS VI including short stature, progressive dysmorphic facial features, hepatosplenomegaly, hearing impairment, corneal clouding, and dysostosis multiplex were largely absent in the younger sibling. Her cardiac functions and joint mobility were well preserved. On the other hand, her affected brother had typical MPS VI phenotypic features described above before commencing ERT at the equivalent age, of 3 years. There was significant improvement in the shoulder range of motion and hearing loss after 120 months of treatment and cardiac function was largely preserved. His skeletal deformity and short stature remained unchanged, but he reached the age of puberty. The results showed that early ERT initiated at newborn is safe and effective in preventing or slowing down disease progression of MPS VI including bone deformities. These observations indicate that early diagnosis and treatment of MPS VI before development of an irreversible disease is critical for optimal clinical outcome..

Biography

Mahoko Furujo is from National Okayama Medical Center, Japan. She has done her graduation from Graduated Hamamatsu University School of Medicine in 1993, Residency in Okayama Medical Center and presently is an attending physician in Okayama Medical Center.

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