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SHORT REPORT

Successful treatment of severe heart failure in an infant with Hurler syndrome

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Summary Hurler syndrome (MPS IH) is the most severe form of mucopolysaccharidosis type I. It is caused by deficiency or absence of the enzyme α-L-iduronidase. Cardiac involvement includes cardiomyopathy and valve and coronary pathology. Cardiomyopathy causing symptoms in an infant with MPS IH carries a very poor prognosis. We describe a previously healthy 10-week-old boy who was admitted to hospital critically ill with severe heart failure. Echocardiography on admission showed severe dilatation of the left ventricle and moderate insufficiency of the left-sided cardiac valves. Accumulation of heparan sulfate and dermatan sulfate substrates in the urine and leukocyte analysis confirmed the diagnosis of MPS IH. Enzyme replacement therapy (ERT) with intravenous laronidase at a standard dosage of 100 U/kg weekly was started soon after. This improved the child's general clinical wellbeing dramatically. His cardiac function improved steadily over a period of months. Stem cell transplantation from cord blood is not available in Norway and he underwent successful transplantation from an unrelated bone marrow donor at the age of 11 months. ERT was stopped four months later. At the age of 26 months his heart function is close to normal and he is currently on no medication. This report highlights three important clinical issues: (1) MPS IH must be considered in infants with cardiomyopathy; (2) early ERT may have a significant impact on short-term outcome in children less than 18 months old with severe cardiomyopathy; (3) our report confirms that patients in poor condition benefit from ERT before stem cell transplantation.

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