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Sommario/riassunto	<p>Pompe disease, also known as acid maltase deficiency or acid alpha-glucosidase deficiency, in its most severe form results in a rapidly progressive, neonatal-onset skeletal and cardiomyopathy, leading to early infantile death without treatment. The development of treatment with recombinant enzyme replacement therapy radically transformed the clinical trajectory of those affected, enabling long-term ventilator-free survival with resolution of cardiomyopathy. These positive clinical outcomes resulted in the implementation of newborn screening programs for Pompe disease across the world. This Special Issue highlights some of the experiences of Pompe screening programs worldwide and discusses public policy and ethical issues elicited by presymptomatic screening for Pompe disease.</p>