

Supplementary

Table S1. Key clinical findings at enzyme replacement therapy start, bortezomib start, and at most recent follow-up at time of database lock for the three patients with infantile Pompe disease.

	Patient 1	Patient 2	Patient 3
Respiratory status at ERT start (week 0)	No respiratory support	No respiratory support	Ventilator dependent
Respiratory status at BTZ start (time since ERT start)	Required ventilation support (PC/PEEP of 15/6 cm H ₂ O, breathing rate increased to 30/min) (weeks 86 – 98)	Required ventilator support (PC/PEEP of 18/8 cm H ₂ O, breathing rate 18/min) (week 146)	Increase in ventilatory requirement ; remained on synchronized intermittent mandatory ventilation without a backup breath rate for 1 hour; increased need for tracheostomy suction (weeks 34-88)
Follow-up respiratory status (time since ERT start)	Decrease in ventilation requirement (PC/PEEP of 15/5 cm H ₂ O, breathing rate 10/min); ability to come off ventilator frequently for short periods (~8–10 min) (week 159)	Decrease in ventilation requirement (PC/PEEP of 10/5 cm H ₂ O, breathing rate 14/min); tolerates trach-mask trials off the ventilator for 10–15 min twice a day (week 221)	Decrease in ventilatory requirements with ability to come off of the ventilator for short periods of time; remains on synchronized intermittent mandatory ventilation without a backup breath rate for the whole day; decreased requirement for tracheostomy suction (weeks 88 onwards)
Gross motor status at ERT start (week 0)	Generalized hypotonia	Generalized hypotonia	Unable to ambulate, sit unsupported
Gross motor status at BTZ start	Voluntary motor activity limited to ocular movements only	Regression of gross motor milestones;	Less energy and less exercise tolerance than previously noted; able to stay in a stander

(time since ERT start)	(weeks 86-98)	remained wheelchair-bound; able to sit with support; unable to stand unassisted (week 146)	for 25 min (week 34 – 88)
Most recent gross motor status (time since ERT start)	Reacquisition of previously lost motor milestones; regained ability to move fingers and toes, bend knees and nod head; recovery of muscles of facial expression and improvement in speech function. Able to operate power wheelchair (week 98 onwards)	Stands with support; increased extremity strength; able to hold arms above body; pushes against resistance (week 200 onwards)	Improved motor control, including holding himself upright while in a stander; able to stay in stander for up to 3 h (week 88 onwards)
Feeding status at ERT start (week 0)	Oral	Oral	Gastrostomy tube
Feeding status at BTZ start	Gastrostomy tube	Nasogastric tube	Gastrostomy tube
Most recent feeding status	Gastrostomy tube	Mostly Gastrostomy tube M&M and hotdogs orally	Oral with improvement in swallow function plus supplementary gastrostomy tube feeds

* ERT, enzyme replacement therapy; BTZ, Bortezomib; PC, pressure control; PEEP, positive end-expiratory pressure