

## Poster Abstract: Therapeutic

### A 4-Year Observational Clinical Study of 24 Adult Pompe Disease Patients under Alglucosidase Alfa Enzyme Replacement Therapy

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#### BACKGROUND

Late-onset Pompe disease is a metabolic myopathy caused by a deficiency of acid alpha glucosidase and is primarily characterised by progressive muscle weakness and loss of respiratory function. Alglucosidase alfa enzyme replacement therapy (ERT) is a licensed treatment for this condition.

#### METHODS

Data were collected retrospectively from 24 patients who received ERT for this disease at our centre. Patients were followed up for an average of 4.3 (range 4–5) years following the start of biweekly intravenous alglucosidase alpha (20 mg/kg body weight). Patients had baseline and follow-up lung function tests, 6-minute walk test, MRC (Medical Research Council) sum score, creatine kinase (CK) levels, SNIP (sniff nasal inspiratory pressure), and quality-of-life SF-36 self-reporting questionnaires. Reliance on ventilation and mobility aid was also assessed.

#### RESULTS

The mean age was  $44 \pm 11.5$  (range 16–64) years at the start of ERT. Follow-up at 2 and 4 years from baseline did not show any significant changes in % predicted forced vital capacity ( $60.2 \pm 26$ , median 55 at baseline and  $58 \pm 24$ , median 56 at 2 years,

$P=0.3879$ ;  $56.8 \pm 22.6$ , median 53 at baseline, and  $58.3 \pm 22.5$ , median 55 at 4 years,  $P=0.2187$ , respectively). At 2 years, the estimated mean changes in SNIP from follow-up at baseline favoured alglucosidase alpha ( $37.36 \pm 20$ , median 32 at baseline and  $48.7 \pm 19$ , median 49 at 2 years,  $P=0.0105$ ), whereas at 4 years from baseline, the estimated mean SNIP did not show any significant change ( $34.25 \pm 22$ , median 27 at baseline and  $40.75 \pm 16$ , median 41.5 at 4 years,  $P=0.1787$ ). The 6-minute walk test between the 2 and 4 years ( $P=0.422$ ) of ERT remained similar. There were significant changes from baseline in the CK levels ( $627 \pm 327$ , median 526 at baseline;  $426 \pm 188$ , median 436 at 2 years;  $P=0.001$  and  $350 \pm 174$ , median 310 at 4 years;  $P=0.0011$ ). At 2 and 4 years, the estimated mean changes in SF-36 mental questionnaire from follow-up at baseline showed no significant change ( $P=0.16$  and  $P=0.26$ , respectively). Similarly, SF-36 physical questionnaire results were not remarkably different at 2 and 4 years from the follow-up at baseline ( $P=0.24$  and  $P=0.47$ , respectively). The reliance on artificial ventilation was 6/24 prior to vs 14/24 patients during ERT. In all, 3/24 vs 4/24 patients were wheelchair bound prior to and during the ERT, respectively.

#### CONCLUSIONS

In this study, cohort treatment with alglucosidase alpha suggests stabilization of pulmonary function over a 4-year period; however, reliance on ventilation and mobility aids increased.

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