Abstract

Objectives Determining the cost-effectiveness of enzyme replacement therapy (ERT) for the classical infantile form of Pompe disease (complete acid α-glucosidase deficiency-related) in two different settings: England and Colombia. Pompe disease is very rare (1:40,000 births incidence). Methods A literature review was made and historic databases searched for National Health Service (NHS) reimbursed costs in England and by health insurers in Colombia; expert opinion was elicited. Two Markov models were constructed for comparing both countries; alive with symptoms and dead were the transition states used. Patients aged 6 months receiving ERT were assumed to have 75% survival rate and better health-related quality of life (HR-QoL) compared to those without treatment (0.700 HR-QoL using the EQ-5D scale). Results The incremental cost-effectiveness ratio (ICER) per quality-adjusted life year (QALY) gained was £234,307.7 for England and £109,991 for Colombia. Uncertainty about final HR-QoL with ERT, disease progression and cost from palliative care had the biggest impact on the ICER in both models. If ERT costs were reduced to 10,000 times per dose and HR-QoL was 0.750-0.820 ICER, then £165,000 could be attainable for England and £65,000 for Colombia. Transaction costs per case in Colombia were high. Conclusions ERT was more effective than no ERT in treating infantile Pompe disease, but high levels of uncertainty still remain about survival and progression rates and QoL in the long-run. ICERs were high compared to CE thresholds. Manufacturers’ ERT costs and monopoly had a major impact on final CEA results.

Keywords

Glycogen storage disease type II, Pompe disease, cost and cost analysis, cost-benefit analysis, quality of life (source: MeSH, NLM).