

Supplementary Information

Clinical Drug Investigation

Cost-effectiveness analysis of newborn screening for spinal muscular atrophy in Italy

Gianni Ghetti¹; Francesco Saverio Mennini^{2,3}; Andrea Marcellusi^{2,3}; Matthias Bischof⁴;
Gabriele Maria Pistillo⁵; Marika Pane^{6,7}

¹AdRes HEOR s.r.l., Turin, Italy; ²CEIS-Economic Evaluation and HTA (EEHTA), Faculty of Economics, University of Rome “Tor Vergata,” Rome, Italy; ³Institute for Leadership and Management in Health, Kingston University London, London, UK; ⁴Novartis Gene Therapies GmbH, Rotkreuz, Switzerland; ⁵Novartis Gene Therapies, Milan, Italy; ⁶Pediatric Neurology Unit, Fondazione Policlinico Universitario “A. Gemelli,” IRCCS, Rome, Italy; ⁷Nemo Clinical Centre, Fondazione Policlinico Universitario “A. Gemelli,” IRCCS, Rome, Italy

Corresponding author:

Gianni Ghetti, MA

AdRes HEOR s.r.l.

Via Vittorio Alfieri, 17

10121 Turin, Italy

E-mail: g.ghetti@adreshe.com

ORCID: 0000-0001-6809-5251

Online Resource Table 1. Clinical aspects of health states in the model [1,2]

State	Motor features	Additional features
A	Within a broad range of normal development	• Within a broad range of normal development
B	Walks unassisted	• No breathing difficulties • Number and severity of chest infections similar to a typically developing child of the same age • Does not require a feeding tube – few difficulties swallowing, is able to eat and, for instance, swallow water • Talking ability similar to that of a typically developing child of the same age
C	Sits unassisted	• May have breathing problems and sometimes require non-invasive ventilation • Development of chest infections more frequently than a typically developing child of the same age • Some difficulties with eating and swallowing but able to swallow thin liquids and take some food by mouth • Risk of choking • Temporary placement of a gastric tube may be required • Requires help moving • Can talk, but ability to speak will deteriorate over time
D	Not sitting	• Experiences breathing problems and requires regular non-invasive ventilation for a number of hours every night or during the day • Development of chest infections more frequently than a typically developing child of the same age • Difficulties feeding and swallowing • High risk of choking • Only able to swallow thick fluids • Fed by a feeding tube (gastrostomy) surgically placed directly into the stomach • Requires moving regularly to prevent sores • Unable to talk, but can make sounds and cry
E	Permanent assisted ventilation	• Requires 24-hour non-invasive ventilation • May require a tracheostomy if non-invasive ventilation is not working well • Requires gastrostomy to be surgically placed directly into the stomach due to difficulty feeding and swallowing • High risk of choking • Requires moving regularly to prevent sores • Development of chest infections more frequently than healthy children of the same age • Unable to talk, but can make sounds and cry

References

1. Velikanova R, van der Schans S, Bischof M, van Olden RW, Postma M, Boersma C. Cost-effectiveness of newborn screening for spinal muscular atrophy in The Netherlands. *Value Health*. 2022;25(10):1696–704. doi: 10.1016/j.jval.2022.06.010.
2. Weidlich D, Servais L, Kausar I, Howells R, Bischof M. Cost-effectiveness of newborn screening for spinal muscular atrophy in England. *Neurol Ther*. 2023;12:1205–20. doi:10.1007/s40120-023-00489-2