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The cost-effectiveness of newborn screening for spinal muscular atrophy and presymptomatic treatment in Estonia

SUMMARY

Objectives: To evaluate the effectiveness and cost-effectiveness of newborn screening (NBS) for spinal muscular atrophy (SMA) and presymptomatic treatment with onasemnogene abeparvovec (OA) or risdiplam compared to risdiplam treatment without SMA NBS.

Methods: A literature review on the effectiveness, safety and cost-effectiveness of presymptomatic treatment with OA or risdiplam was composed. Cost-effectiveness analysis with a time horizon of 5, 10 and 20 years was conducted by combining a decision tree and a Markov model. A decision tree was designed to capture the initial NBS outcomes and treatment options. Subsequently, a Markov model was linked to simulate the health outcomes and costs. The model transitions for presymptomatic treatment with OA were derived from the SPR1NT trial. As risdiplam is currently under investigation for newborns with presymptomatic SMA, the same treatment efficacy for OA treatment in presymptomatic SMA was applied. Quality of life estimates were derived from published literature. Costs from the societal perspective included costs of screening (including true and false positives), diagnosis, disease-modifying therapies, drugs, direct medical care and parents' loss of productivity to care for SMA children. Costs and QALYs were discounted using an annual discount rate of 5%. Results were presented in terms of costs, quality adjusted life-years (QALY) and incremental cost-effectiveness ratios (ICER). A 5-year budget-impact analysis was carried out from the healthcare payer perspective.

Results: In the base case scenario, the analysis showed that NBS for SMA and presymptomatic treatment with OA would enable to gain 2.44, 4.42 and 7.17 QALYs in the perspective of 5, 10 and 20 years, respectively, compared to the risdiplam treatment without SMA NBS. Respective ICERs were estimated at €1,116,457, €484,065 and €183,674 per QALY gained. In the additional hypothetical scenario, compared to no NBS and treatment with risdiplam, NBS and presymptomatic risdiplam treatment resulted in ICERs of €111,904, €50,047 and €20,667 per QALY gained over 5, 10 and 20 years, respectively. The results were most sensitive to the proportion of SMA positives found in the screening, prevalence rate of SMA I–III in Estonia, the price of OA and non-application of discounting. According to the budget impact analysis, the additional cost of NBS for SMA and presymptomatic treatment with OA would be 2.72 – 3.14 million euros in year compared to the risdiplam treatment without SMA NBS. The additional annual cost of NBS for SMA and presymptomatic treatment with risdiplam would be €209,000 – €297,000.

Conclusions: SMA NBS with presymptomatic treatment with OA or risdiplam improves the quality and length of life for infants with SMA, but treatment with risdiplam is associated with lower costs than OA treatment.

Citation: Juus E, Õunap K, Kahre T, Reinson K, Lutsar K, Sarv S, Müürsepp M, Jürisson M. *Spinaalse lihasatroofia sõeluuringu ja presümptoomse ravi efektiivsus ja kulutõhusus.* TTH59. Tartu Ülikooli peremeditsiini ja rahvatervishoiu instituut; 2022.