The cost-effectiveness of newborn screening for cystic fibrosis in Estonia

SUMMARY

Objectives: To evaluate the effectiveness and cost-effectiveness of adding newborn screening (NBS) for cystic fibrosis (CF) to an existing neonatal screening programme in Estonia.

Methods: To meet the objective, a literature review of the effectiveness and cost-effectiveness of NBS for CF was composed. Estonian cost-effectiveness calculations with a time horizon of 80 years were conducted by combining a decision tree and a Markov model. The screening algorithm consisted of measuring the level of immunoreactive trypsinogen (IRT), then determining the pancreatitis associated protein (PAP) level and a test of deoxyribonucleic acid (DNA) was performed in the third stage. The cost-effectiveness of two NBS algorithms were separately assessed, the difference was in the method of DNA analysis – Sanger or whole CF transmembrane conductance regulator gene (*CFTR*) sequencing. The model transitions for NBS were derived from the Estonian data and the treatment efficacy for CF was based on the public literature. Quality of life estimates were also derived from published literature. Screening, drug and treatment costs were calculated using Estonian Health Insurance Fund (EHIF) data, whose perspective the analysis employed. 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 implementing NBS for CF could result in gains of 1.16 and 1.27 QALYs through DNA Sanger sequencing and whole *CFTR* gene sequencing, respectively, compared to no screening. Respective ICERs were estimated at €37,997 – 117,199 and €71,403 – €151,573 per QALY gained. The cost-effectiveness ratios were most influenced by the proportions of positive IRT, PAP, and DNA tests, quality of life estimates for CF, and the cost of supportive treatment for CF without NBS and with Kaftrio + Kalydeco treatment. According to the budget impact analysis, the additional cost of NBS for CF would be €540,600 – €544,300 euros and €668,400 – €672,300 euros over five years through DNA Sanger sequencing and whole *CFTR* gene sequencing, respectively, compared to no NBS.

Conclusions: The NBS for CF by using whole *CFTR* gene sequencing is more effective compared to the DNA Sanger sequencing. Additionally, whole *CFTR* gene sequencing identifies one more newborn with CF every 7–8 years compared to DNA Sanger sequencing.

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