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Cost-effectiveness analysis of Next generation sequencing tests in critically ill pediatric patients

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Rare genetic diseases in the pediatric population constitute an urgent global public health issue; overall, more than 300 million people are affected worldwide. Next Generation Sequencing techniques, as Whole genome sequencing (WGS) and Whole exome sequencing (WES), have proven to be significantly supportive in diagnosing these complex conditions. The aim is to evaluate the cost-effectiveness of WGS versus WES in pediatric patients with suspected genetic disorders. A Bayesian Markov model was calibrated among this target population, comparing WGS to WES. Model parameters were retrieved from the scientific literature. Costs and benefits were discounted at a rate of 3%. A lifetime time horizon and the National Health Service perspective was chosen. The Eurozone threshold, ranging from €30,000 to €50,000, was adopted. Markov Chain Monte Carlo was used as the simulation method for Bayesian inference. Uncertainty was explored through a probabilistic sensitivity analysis (PSA) and a value of information analysis (VOI), illustrated through Cost-Effectiveness Acceptability Curve (CEAC) and Expected Value of Perfect Information (EVPI). Results were reported as Incremental Cost-Effectiveness Ratio (ICER), expressed as euros per additional diagnosis. The base case findings highlighted that WGS was cost-effective with an ICER of €31,973. The CEAC showed that for all thresholds over the ICER, WGS had the highest probability of being cost-effective. The EVPI per patient was estimated to be €6,535 on a threshold of €50,000/diagnosis. In addition to being cost-effective, WGS could allow early genetic diagnosis shortening the diagnostic odyssey. The use of WGS in the diagnostic workup has the potential to revolutionise personalised medicine and to play a significant role in achieving SDG 3 by providing personalised healthcare, identifying genetic risk factors for diseases, and informing public health policies for a target population that represents the human capital of the future.

Key messages:

- This analysis informs that WGS is cost-effective and possibly cost-saving in the long term.
- WGS could be implemented in the diagnostic workup of children with suspected rare disorders.