The Hospital for Sick Children
Technology Assessment at SickKids (TASK)

EXECUTIVE SUMMARY

MICROCOSTING OF WHOLE GENOME SEQUENCING (WGS) OF TRIOS IN A HETEROGENEOUS PEDIATRIC CARDIAC POPULATION

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Executive Summary

Background

Multiple causes of heart failure (HF) in children and the challenges in treatment and management of care has increased demand for whole genome sequencing (WGS). WGS captures information that could help determine the cause of or risk factors associated with HF not only for patients but also for family members. Identifying a genetic cause or risk factor can, in turn, aid in clinical decisions related to screening, treatment and management. An economic evaluation of WGS technology requires a comprehensive and accurate estimation of all costs involved in the sequencing workflow. This would aid in policy and implementation decisions of this technology for the pediatric HF patient population.

Objectives

The objective of this study was to estimate costs per trio for WGS, including coding and non-coding regions, for a targeted patient population consisting of children with heterogeneous cardiac diseases including cardiomyopathies (CMP), congenital heart defects (CHD) and inherited cardiac arrhythmias enrolled in the cardiac genome clinic (CGC) at The Hospital for Sick Children (SickKids), Toronto, Canada from an institutional payer perspective over five years.

Methods

Using a bottom-up microcosting approach, the opportunity cost per trio excluding mark-ups, fees and charges for WGS-trios on the Illumina HiSeq X™ platform for pediatric patients with multiple cardiac diseases was estimated. This was done from an institutional payer perspective based on the diagnostic laboratory practices at SickKids. The cost per trio was determined for each year of a five-year program. Total program costs to service the CGC pediatric population were also estimated over five years. A probabilistic analysis (PA) was conducted to incorporate parameter uncertainty in the model. Three one-way deterministic sensitivity analyses (DSA) were conducted to examine the effects of changing the inputs for the overhead cost, the total volume of WGS tests in the institution, and excluding pharmacogenomics while other inputs remained the same.

Results

The cost per trio in Year 1 was $8053 (95% confidence interval [CI]: 7699, 8558) for WGS-trio (HiSeq X™). Reagent supply costs accounted for the largest proportion of costs (50%) followed by bioinformatics

(25%). The total institutional program cost to offer WGS for CGC diagnosis over five years was $5.63 million (95% CI: 5.38, 5.98) based on 144 CGC trios per year. Varying the inputs in DSAs resulted in a minimal difference of under 5% in the overall costs per WGS-trio.

Conclusions

This study estimated the cost of trio WGS using a bottom-up microcosting approach. The study provides comprehensive cost data for use in future economic evaluations of genome sequencing in pediatric cardiac patients. It allows for a costing model that can be easily updated as technology evolves and adapted to other pediatric patient populations. Additional analyses are required to assess the clinical and economic impact of the WGS in this population.