OP168 Costs And Effectiveness Of Whole Exome Sequencing (WES) In Patients With Unsolved Rare Disease Through The Diagnostic Pathway

Deborah A Marshall (damarsha@ucalgary.ca), Koen Degeling, Toni Tagimacruz, Trevor A. Seeger, Kym M Boycott, Francois Bernier, Roberto Mendoza-Londona, Karen V. MacDonald, Taila Hartley and Robin Z. Hayeems

Introduction: Patients suspected of having a rare genetic disease often experience lengthy and costly diagnostic odysseys. The timing of whole exome sequencing (WES) in the testing sequence, its diagnostic yield and test costs in the sequence all factor into estimates of cost-effectiveness analysis for health technology assessment.
Methods: We modeled the diagnostic pathway using a discrete event simulation model, starting with the first test result. We defined and populated the simulation based on data from the electronic medical records of n=307 from the Care-for-Rare SOLVE multi-center Canadian observational cohort. Five alternative diagnostic pathways were modeled based on the observed data: no WES, and WES as the first, second, third or fourth test in the sequence. WES as the second test in the sequence is considered standard of care in medical genetic centers in Canada. We assessed effectiveness of WES in terms of diagnostic yield, time to diagnosis, and costs as patient-level overall test costs (2020 CAD/USD) across the diagnostic pathway.
Results: Compared to molecular and specialized diagnostic tests only (i.e., no WES), WES increased diagnostic yield from 5 percent to 40 percent. The shortest time to diagnosis for those with a diagnosis was 1.82 years in the diagnostic pathway with WES as the second test. Test costs for each pathway were CAD2,800 (USD2,087, no WES), CAD2,700 (USD2,013, WES as first test), CAD3,500 (USD2,609, WES as second test), CAD4,500 (USD3,354, WES as third test), and CAD5,300 (USD3,951, WES as fourth test).
Conclusions: Placing WES earlier in the diagnostic pathway for patients suspected of having a rare disease is associated with an increased diagnostic yield, reduced time to diagnosis and lower overall test costs with the benefits being greater the earlier in the pathway that WES is implemented.

OP171 Canadian Disease Registry Inventory: Environmental Scan Of The Literature

Amanda Hodgson (amandah@cadth.ca), Hannah Loshak, James Lachaud, Amélie Bernard, Teri Slade, Geneviève Mák and Heather Smith Fowler

Introduction: In consideration of the lessons learned from other jurisdictions and other ongoing work in the disease registry data space, an opportunity existed to investigate the current Canadian landscape and identify opportunities for a Canadian registry list. Previously, no national-level inventory of registries existed in Canada that could provide the necessary information to support awareness and use of available data for decision-making.
Methods: A literature search was conducted on key resources, including MEDLINE and a focused internet scan. No methodological filters were applied to limit retrieval by publication type. The search was limited to documents published in English or French.
Results: Core characteristics of the identified registries were extracted and contextual information on the current landscape of disease registries in Canada was explored. A literature review and draft inventory list has been produced.
Conclusions: A CADTH environmental scan was undertaken to collect and report on existing Canadian disease registries and to identify key features, characteristics, and intersections. This information and analysis increase the potential of Canadian registries to inform decision-making and identifies opportunities for the optimal use of registry data in Canada more broadly.

OP172 International Collaboration For Translating The Peer Review Of Electronic Search Strategies (PRESS) Checklist: A Harmonized Approach

Amanda Hodgson, Daniele Masterson, T. P. Ferreira, Josué Laguardia, Cicera Henrique da Silva, Martha Silvia Martinez Silveira, Jessie McGowan and Danielle Rabb (danieller@cadth.ca)

Introduction: The PRESS guideline and checklist provides a set of recommendations concerning the information that should be used by

https://doi.org/10.1017/S0266462323001630 Published online by Cambridge University Press

librarians and other information specialists when they are asked to evaluate electronic search strategies. CADTH and PRESS authors were approached for permission to translate this checklist into various languages.
Methods: The team from Fundação Oswaldo Cruz and the Universidade Federal do Rio de Janeiro asked CADTH for permission to translate the PRESS guideline and checklist for their research work. They translated PRESS following the steps advocated by the scientific literature on the translation of standardized questionnaires.
Results: CADTH is now sharing and actively disseminating the PRESS translations in French and Portuguese (and a forthcoming version in Spanish) via the Finding the Evidence website and through related presentation activities.
Conclusions: The coordinated translation of key health technology assessment (HTA) tools provides an avenue for international uptake and improvement of best practice in information retrieval, which is a foundational feature of HTA work. With the absence of formal translation guidelines on the translation of protocols such as PRESS, CADTH would benefit from developing guidance for HTA teams requesting to translate our tools. CADTH is currently conducting research on the uptake and use of PRESS, which will inform future knowledge mobilization strategies such as translation standards and communications.

OP173 Estimating The Marginal Productivity Of Health Technology Adoption

Charles Yan (cyan@ihe.ca), Jemal Mohamed, Manik Saini, Robert Bacigalupo, Selva Bayat and Jeff Round

Introduction: Decisions to adopt health technologies rely, in part, on judgements about cost effectiveness. Cost effectiveness is commonly assessed against a willingness-to-pay threshold for health gains. Building an evidence base on the marginal productivity of health spending to inform the value of the threshold is increasingly of interest for resource allocation decision-making and technology implementation. We report on an in-progress analysis to inform a threshold for policy purposes in British Columbia, Canada.
Methods: We developed a ten-year panel-data model with instrumental variables, which lessens the degree of time-invariant confounding and addresses biased causal inferences caused by unobserved factors, to provide estimates of the marginal cost per health unit measured using quality-adjusted life-years (QALYs). We use the Johns Hopkins Adjusted Clinical Group (ACG) system and a British Columbia Health System Matrix to classify patients into six resource use bands (RUBs) ranging from 'healthy' to 'very high morbidity'. Patients are also classified by chronic conditions and types of services. Place of residence and geographical region of health authorities are considered. Variables included age, gender, mortality and comorbidity rates, costs of hospitalizations, emergency department and physician visits, residential and home care, laboratory services, diagnosis and medications, and quality of life. Instrumental variables included sociodemographic characteristics as reported in the Canadian census.
Results: The largest RUB was 'moderate' morbidity (39.3%), while the smallest was 'healthy' (1.5%). The youngest was the 'low' morbidity (mean 31, standard deviation [SD] 21) and the oldest was 'very high' (mean 69, SD 17). The healthy group had the smallest mean costs (CND563, SD CND4,121; equivalent to USD421, SD USD3,083). In contrast, the 'very high' group had the largest (CND20,398, SD CND36,188; equivalent to USD15,258, SD USD27,069). Age and gender standardized comorbidity index scores ranged from 0.05 to 6.41 (median 0.98). Additional analyses (e.g., costs per QALY) are ongoing and the results will be reported at the conference.
Conclusions: Our empirical approach is robust and flexible, allowing estimates of marginal productivity according to factors such as disease, geographical region, service type, and care sector. This work has applications at the provincial and national levels and adds to methodological literature in the field.

OP174 Health Technology Assessment And Economic Evaluations For A Genomic Strategy In Italy

Eugenio Di Brino (eugenio.dibrino@unicatt.it), Debora Antonini, Giulia Falasca, Michele Basile, Filippo Rumi and Americo Cicchetti

Introduction: The challenge to health systems is sustainability, not only in the economic and financial sense of compatibility of spending with allocated resources, but also in terms of equity in access to services and care, quality, safety, innovation and research, that is, in terms of the effectiveness of the right to health. Some countries, such as Italy, do not have formal health technology assessment (HTA) or other similar processes that take into account views outside the decision maker.
Methods: In the Italian national context, there is currently no single tariff for the reimbursability and pricing of genomic technologies. In fact, although genomic tests have been in clinical practice for many years now, to date they have not yet been included in the LEA (Minimum healthcare provision), especially in view of the fact that a defined and transparent process for updating the Essential Levels of Care has been operational since 2018. With the goal of structuring guidelines for the adaptability of economic evaluations to currently available and developing genomic technologies, a literature review was conducted.
Results: The literature review showed that there are some methodological and practical issues that need to be carefully considered when designing and conducting economic evaluations of genomic tests. In more detail, five key concepts were identified in order to implement the most comprehensive economic evaluation of the technologies under study: the PICO model, the survey perspective, the costs included in the analysis, the effectiveness analysis, the time horizon, and the discount rate.
Conclusions: Adequate definition of these concepts appears to be of paramount importance in view of the fact that genomic testing may

https://doi.org/10.1017/S0266462323001630 Published online by Cambridge University Press