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

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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

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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

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

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