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DRUG APPROVAL FOR RARE METABOLIC DISEASES- SHOULD REAL WORLD DATA FROM GLOBAL PATIENT REGISTRIES CONSTITUTE EVIDENCE?

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Background:
The natural history of untreatable rare genetic disorders is changing with the emergence of more and more effective treatments. The drug approval process in Canada is complex with many layers. Each disorder is very rare and thus there is a limited number of patients available for clinical trials to generate the evidence needed to prove efficacy and safety. We present our experience with 3 patient Registries and the potential that their "real world data" (RWD) can play an important role in the drug approval process and inform treatment guidelines.

Objective:
To review our recent experience with 3 patient Registries (2 Industry-sponsored and 1 Investigator-led) for select hereditary metabolic disorders.

Methods:
Publications on RWD using PubMed were reviewed from 2 global Industry-sponsored Registries -[Sanofi Genzyme Registry NXT for 4 lysosomal storage diseases including Fabry disease (FD) and Alexion Hypophosphatasia (HPP) Global Patient Registry] - and 1 Investigator-led Canadian Fabry Disease Initiative – National Registry (CFDI-NR). Inputted data on enrolled FD and HPP patients were summarized and compared with published Registry data.

Results:
Longitudinal data on our 17 FD and 25 HPP consenting patients represent <1 % and 3.7 % of patients enrolled in their respective Global Registries and 4.8 % of FD pts in the CFDI-NR. RWD inputted include Quality of Life (Q of L) questionnaires, Depression scale, clinical/laboratory results, and safety data in both ERT treated and untreated patients. Helpful treatment and monitoring recommendations have resulted from Registry data.

Conclusion:
RWD, particularly on Q of L and burden of disease, can capture important information sometimes missing from clinical trial data. These data help inform clinical practice and develop important research questions needing to be explored. Limitations of such data include incomplete or overlapping data sets, bias of industry-sponsorship, and the absence of a National Canadian Registry framework to enhance efficiency, ensure transparency and equity.