

Establishing a comprehensive analytical framework for these unique diseases poses difficulties. This study aims to descriptively analyze arguments reported by the Brazilian National Committee for Health Technology Incorporation (Conitec) in deciding whether to include technology for ultra-rare diseases.

Methods: Data from recommendation reports (2012 to 2022) were analyzed. Diseases with a prevalence of fewer than one per 50,000 inhabitants were classified as ultra-rare. Extracted variables included preliminary and final recommendation results and justifications by Conitec. Six argument categories were created (method-related issues; evidence; cost; technology effectiveness or safety; context; innovation). Word clouds were generated based on word frequency in each category to present the data.

Results: In the analysis of 45 reports, the word clouds highlight frequent terms in favorable arguments, emphasizing evidence quality, cost reduction, and applicability in the healthcare system. Conversely, unfavorable arguments also revolve around evidence quality and cost impact. The analysis of the arguments according to categories, 16 arguments were identified: seven concern evidence issues, five cite methodological problems in presented studies, four relate to costs, and three pertain to technology effectiveness or safety. Unfavorable arguments primarily stem from evidence-related concerns. In favorable arguments, cost (seven) and safety (six) are prominent, with innovation (one) and context (three) being additional categories not found in the unfavorable group.

Conclusions: While technology assessment processes for ultra-rare diseases have evolved, the justifications for recommending or not incorporating new technologies remain unchanged. Over time, reports have become more detailed, focusing on evidence and methodological specifics. This highlights the importance of scrutinizing evidence characteristics and determining relevant criteria and data types for this unique context.

PP29 Social Preferences In Health Technology Assessments For Rare Diseases: A Systematic Literature Review Of New Analytic Approaches

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Introduction: Rare diseases (RD) can be severe and can dramatically reduce life expectancy and quality of life. RD therapies, mostly orphan drugs (OD), fail to meet the standard criteria for public reimbursement due to uncertainty in cost-effectiveness estimates limiting healthcare access. To address this issue, experts have suggested the integration of social preferences into health technology assessments (HTAs) by implementing different methods.

Methods: This systematic literature review, performed in 2021, aimed to explore worldwide experiences of social preferences integration into HTAs for RD and OD through the implementation of multiple-criteria decision analysis (MCDA), discrete choice

experiments (DCE), and person trade-off (PTO), among other methods. A systematic search of the literature was conducted using PubMed, Cochrane, Embase, and Scopus databases. The PRISMA approach was used for the review phases. Finally, the Promoting Action on Research Implementation in Health Services (PARIHS) framework was used to discuss the implementation of these instruments in the RD context.

Results: Thirty-three articles met the inclusion criteria. The studies measured social preferences for RD and OD as part of HTA using MCDA (n=17), DCE (n=8), and PTO (n=4), among other methods (n=4). These found that patients and clinicians do not prioritize funding based on rarity. The public is willing to allocate funds only if OD demonstrates effectiveness and improves the quality of life (QoL), considering as relevant factors disease severity, unmet needs, and QoL. Conversely, HTA agency experts preferred their current approach and placed more weight on cost-effectiveness and evidence quality even though they expressed concern about the fairness of the drug review process.

Conclusions: MCDA, PTO, and DCE, among others, are helpful and transparent methods for assessing social preferences in HTAs for RD and OD. However, their methodological limitations, such as arbitrary criteria selection, subjective scoring methods, framing effects, weighting adaptation, and value measurement models, could be hurdles to implementation. Further research is needed to tailor these methods' applicability and impact in different social contexts.

PP30 The Health System Impact Of Returning Rare Disease Variants As Secondary Findings From Genomic Sequencing: A Population-Based Model

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Introduction: The American College of Medical Genetics and Genomics (ACMG) recommends that pathogenic variants linked to 37 genetic diseases be disclosed as secondary findings (SFs) to patients undergoing genome-wide sequencing (GWS), including rare diseases (RDs) treated with enzyme replacement or gene silencing/replacement therapies. We estimated the potential budget impact of treating these RD patients in the US and Canada.

Methods: A population-based model was used to estimate the number of patients that would be identified in a given jurisdiction for the 11 diseases on the ACMG's SFv3.2 list classified as "inborn errors of metabolism" or "miscellaneous phenotypes." Genetic and clinically diagnosed prevalence was estimated for each gene-phenotype pair based on the scientific literature. Demographic and GWS utilization data were obtained from government websites and payer reports. Drug costs were obtained from the literature and payer websites and reported in 2023 US dollars. A range of one-way sensitivity and