Values and preferences for the development of a funding model for rare and rare undiagnosed diseases

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Background

Most rare diseases are genetic disorders leading to congenital anomalies, patients with intellectual disabilities, an altered immune system, neurological disorders among others, all of which are a significant cause of premature mortality, morbidity and disability [1]. Although they are jointly classified as rare diseases, they are heterogeneous and complex in nature, their low frequency makes them difficult to recognise and treatments are extremely limited. All of these characteristics make rare diseases a challenging problem, not only from a clinical but also from a health care system perspective.

From a policy perspective, there is no clear pathway or recommendation regarding how rare diseases should be handled. In most cases the management and/or treatment is extremely costly making this topic a focus of considerable policy attention. Under the current model of research and development of new drugs, there is little incentive for pharmaceutical companies to develop new drugs given the high cost associated to bringing a new drug to the market and the challenges in recovering the investment given the small target population [2]. Before even thinking about a potential treatment, a patient needs to be first diagnosed, which is another major challenge that people with rare diseases face. The rarity of the conditions means a lack of familiarity with treating these diseases, thus it is very common that patients experience substantial delays in reaching a correct diagnosis and adequate care. Studies in Mexico, Canada and Europe, showed that a quarter of participants had a 5-30-year delay in reaching a correct diagnosis and may have consulted over 20 specialists in the process. Moreover, close to half received erroneous diagnosis and therefore, inappropriate health care provided [3]. The consequences of health systems not prioritizing rare undiagnosed diseases in terms of morbidity, mortality, quality of life and overall direct and indirect costs, have not yet been quantified [4-6]. Rare undiagnosed diseases remain invisible, patients that suffer them face substantial barriers in healthcare and are generally managed with suboptimal treatments.

Vision

Funding treatments for rare diseases is a global problem. Small patient numbers and high drug development costs, means that there are limited incentives for private industry to develop new treatment options, and the high treatments costs are beyond the means of most patients, potentially leading to inequitable access to treatment. This market failure necessitates the need for Government funding, which generally lies outside the value for money framework. Understanding patient's, carers and other stakeholder's preferences regarding funding models is critical in order to define a value framework on rare diseases, where attributes and opportunity costs are presented transparently enhancing more legitimate decision-making process.

Aims and objectives.

The main aim of this project is to determine the value and preferences of individuals (patients, carers and/or people in the community) that will be used to develop a value framework for rare and rare undiagnosed diseases to support a funding model and ultimately, a decision-making process.

To achieve this, I will need to:

1. Review the funding models implemented worldwide targeting rare diseases and identify the different attributes considered in their development.
2. Evaluate the preferences of the society/patients/carers towards health technologies (treatments and diagnostic tools) that target this patient population.

Research methodology

Methods

A systematic literature review will be conducted to identify all possible funding arrangements implemented worldwide regarding the reimbursement of health technologies targeting patients with rare diseases. The literature search will review other literature reviews, guidelines and policy articles. A targeted survey directed to specific HTA agencies with experience in the assessment of rare diseases may also be conducted to identify more recent approaches that may not yet be available in the published literature.

Discrete choice experiments are stated preference methods to assess complex multi-attribute services when limited data are available. A DCE will be conducted with the aim of generating the required data by eliciting stated preferences from individuals [7]. The systematic literature search will frame the question that the DCE will aim to answer. For example, to understand the preferences between an intervention targeting a rare disease (i.e. high cost, disabling and affecting young children) versus an intervention that targets a life threatening condition (i.e. high cost, high mortality and affecting adults). A question we may wish to answer through a DCE will be whether the society is more willing to fund a very expensive drug to a child which will lead to an additional 10 years of life with a relatively poor quality of life or fund a drug that will save a life to and adult that will live his life expectancy with a good quality of life. Similarly, we may be interested in assessing the preferences of the society/patients/carers towards technologies that improve the rate of diagnosis of patients that will otherwise remain invisible to the health system.

The table below presents each specific objective, its corresponding methodology and the data that will be used accordingly.

Impact to clinical genomic implementation

The value and preferences of relevant stakeholders regarding technologies that may improve the diagnose (through advanced genetic testing) of rare diseases is currently unknown. Moreover, there is no funding model that incorporates the preferences of patients/carers and people in the community in a systematic and transparent way to ensure access to timely diagnose through genetic testing and subsequent treatment of rare diseases. In general, existing funding models focus on technologies

targeting treatment and not in diagnostic tools (like genetic testing) where the consequence are patients that remain invisible to the health system.

Collaborators (other organisations)

This research proposal is part of a larger international research collaboration being conducted in Chile which was funded by the 'NATIONAL RESEARCH FUNDING COMPETITION FONDECYT 2021'. This research proposal is also part of a PhD scholarship application at the Centre for Health Economics and Research Evaluation (CHERE) of the University of Technology Sydney, Australia.

Future funding

Other activities related to this research will be conducted through a PHD scholarsh from the University of Technology Sydney.

Research plan

	Q1	**Q2**	**Q3**	**Q4**
Research activities	Systematic literature review	Identification of attributes and	DCE design and implementation	Data analysis
Goal	Review the funding models implemented worldwide targeting rare diseases and identify the different attributes considered in their development.	Evaluate the preferences of the society/patients/carers towards health technologies (treatments and diagnostic tools) that target this patient population.		

References

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Budget

The values presented in the table below are reference prices only as no quotes have been formally requested. The total cost will depend on the DCE design and number of surveys needed. The latter will be derived from the results in Q1-Q2.

The resources required are needed to pay for the data collection. The analysis will be conducted as part of the usual research activities.

DCE Quotes (PureProfile)		
Survey Construction		7,000
Survey Hosting		1,000
Survey Recruitment		11,000
Total		**US 18,000**
Travel and meeting support		
Conference presentation (likely in Chile)		US 700
Tickets (Sydney/Santiago)		US 2,700
Accommodation		Not required
Total		**US 3,400**
Total required		**US 21,400**

Note: Pureprofile is the company that provides the data collection platform to conduct the DCE. This is the company that CHERE at UTS is most familiar with.