METHODS:

A targeted literature review and social media listening project guided design of a discussion guide for in-depth patient interviews (n=12). These, in turn, guided construction of a quantitative questionnaire administered to moderate to severe DED patients, 40 per country in Australia, Germany, United States and United Kingdom (total n=160). Patients' preference structures were explored through an online survey using a self-explicated conjoint methodology, because of its high respondent-friendliness. Additionally, we administered the EQ5D-5L instrument to determine the health states/utilities of patients. Reaction to a hypothetical novel treatment was further obtained to check for convergent validity with the self-explicated conjoint. Finally, we asked respondents to rate the ease and relevance of the questionnaire to them.

RESULTS:

Qualitative research uncovered important patient perspectives that were built into the quantitative survey. For example, patients seek medical advice when their symptoms are not improving. Patients' lives are most affected by sensitivity to light, itchy and tired eyes and an inability to perform computer/screen work; however, of most concern/worrying to them is that their DED will get worse and they go blind. Results from the quantitative preference research will also be shared and its implications for future clinical trials in DED outlined. The results of the patient research and preference study are to be shared with health technology assessment (HTA) bodies and regulators through the early dialogue scientific advice process.

CONCLUSIONS:

A process of using qualitative research to determine what matters to patients and then quantification through respondent-friendly preference research can identify outcomes that are most patient-relevant, to inform future drug development strategies.

PP48 Caregiver Perceptions And Experiences Of Diagnostic Genome-Wide Sequencing

AUTHORS:

Jenny Hong (jenny.hong@alumni.ubc.ca), Annalijn Conklin, Nick Dragojlovic, Alison Elliott, Jan Friedman, Larry Lynd

INTRODUCTION:

The objective of this qualitative analysis was to explore caregiver perceptions and experiences of receiving or not receiving a genetic diagnosis following genome-wide sequencing (GWS) in children with suspected genetic disorders.

METHODS:

One caregiver for each child completed an online survey two weeks after enrolling in the Clinical Assessment of the Utility of Sequencing and Evaluation as a Service (CAUSES) study, beginning in January 2016, and again about six months after receiving the GWS results. The survey covered the caregivers' experiences and quality of life and children's healthcare resource utilization, and provided open-ended questions for comments. The follow-up survey was completed by twenty families who had received a diagnosis with their GWS results and by twenty-two families who had not received a diagnosis. A thematic analysis of the free-text comments from both groups was performed using NVivo 11.4.2.

RESULTS:

Caregivers from both groups expressed similar experiences of negative socioeconomic effects of caregiving, particularly related to employment and time burden. Caregivers who did not receive a diagnosis with the GWS results were generally hopeful of receiving a diagnosis in the future and reported expectations of a positive benefit from receiving a diagnosis, both in terms of access to additional resources and of positive psychological effects. The absence of a diagnosis was a source of anxiety for many caregivers. By contrast, caregivers who had received a diagnosis reported positive, neutral, and negative psychological effects from the knowledge gained; no participants commented on the consequences for access to additional services or other socioeconomic effects.

CONCLUSIONS:

Our findings suggest that caregivers may have high expectations for what a diagnosis can provide to them and their families, which may not be fully met once a diagnosis is obtained. The study underpins the importance of patient-centered communication of genomic testing results so that families can set realistic expectations of what having a diagnosis will achieve.

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