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Acceptability and feasibility of an online information linker service for caregivers who have a child with genetic epilepsy: a mixed-method pilot study protocol

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ABSTRACT

Introduction Developmental and epileptic encephalopathies (DEEs) are rare epilepsy conditions that collectively impact 1 in 2000 children. They are highly genetically heterogeneous, resulting in significant barriers to accurate and adequate information for caregivers. This can lead to increased distress and dissatisfaction with the healthcare system. To address this gap, we developed 'GenE Compass' to provide caregivers with the highest-quality possible, understandable and relevant information in response to specific questions about their child's DEE. Using a mixed-method design, we will now pilot GenE Compass to evaluate the acceptability to caregivers and clinicians, feasibility and impact to caregivers.

Methods and analysis We will recruit 88 caregivers (estimated final sample of 50 at follow-up) who have a child under 18 years of age with a suspected or confirmed DEE diagnosis. Following consent and a baseline questionnaire (questionnaire 1 (Q1)), participants will be able to submit questions to GenE Compass over a 3-month period. After 3 months, participants will complete a follow-up questionnaire (Q2) and an optional telephone interview to answer the research questions. Primary outcomes are acceptability of GenE Compass and feasibility of delivering the intervention (eg, cost of the intervention, number of questions submitted and time taken to respond to questions). Secondary outcomes include the impact of GenE Compass on caregivers' quality of life, information searching behaviours, perceptions of their child's illness and activation.

Ethics and discussion The study protocol (V.2, dated 16 September 2021) has been approved by the Sydney Children's Hospitals Network Human Research Ethics Committee (ETH11277). The results will be disseminated in peer-reviewed journals and at scientific conferences. A lay summary will be disseminated to all participants.

Trial registration number ACTRN12621001544864.

STRENGTHS AND LIMITATIONS OF THIS STUDY

⇒ A strength of our study design is the mixed-method approach, with interview data used to complement our quantitative survey findings.
⇒ A strength of our study is the substantial consumer involvement in the protocol design, participant documents and questionnaires.
⇒ Our study is limited due to the exclusion of the potentially most vulnerable groups—culturally and linguistically diverse families, and those who are experiencing acute distress.
⇒ We have allowed for 3 months' access to GenE Compass; however, this may not be enough time for caregivers to submit their questions and/or receive benefit from the reports we have prepared.

INTRODUCTION

Rare disease is a major public health challenge. There are over 10 000 rare conditions that affect an estimated 8% of the population,[1] many of which have their onset in childhood and a genetic cause. In December 2021, the United Nations (UN) adopted the first ever UN 'Resolution on Addressing the Challenges of Persons Living with a Rare Disease and Their Families'[2] as part of a global movement to improve care for people living with rare diseases. The Australian government also released a National Strategic Action Plan for Rare Diseases in 2020,[3] highlighting the importance of improved rare disease awareness, support, management and research.

One rare disease cohort requiring further support is children affected with a developmental and epileptic encephalopathy (DEE). DEEs are characterised by childhood-onset drug-resistant seizures and developmental slowing or regression.[4] Children with DEE have a high mortality and a complex range of comorbidities such as autism spectrum disorder, motor deficits and sleep disorders.[4]

As with many rare diseases, these complex care issues lead to recurrent hospitalisations, sudden risk of death and high burden of care. Although DEEs are collectively common with an incidence of 1 in 2000,[5] they are individually ultra-rare,[6] with each individual DEE affecting an estimated less than 1 per 2 000 000 people. This is because DEEs are genetically heterogeneous with over 400 monogenic causes.[7] Many genetic causes have only recently been identified due to significant advancements in genomic sequencing.[7 8] Even when a genetic cause is identified, this can have a vast range of comorbidities and phenotypes.

These factors mean that there is very little information regarding natural history, prognosis and comorbidities, and limited dedicated patient advocacy or support services for DEE,[9] a common theme across rare disease care.[3 8 10] Of the limited information about DEE, the frequently generic nature of this information often limits relevance to the unique situation of each child.[11]

Understanding their child's condition is critical for caregivers. Unmet information needs and limited psychosocial resources can contribute to greater levels of stress and poorer psychosocial outcomes.[11 12] This is indeed seen in caregivers of a child with DEE who report higher levels of anxiety and depression than population norms,[13–18] and in comparison to caregivers of other chronic childhood-onset conditions such as cerebral palsy and Rett syndrome.[19] Caregivers have identified that barriers to information impede access to appropriate and timely healthcare, education and community services.[11] They are desperate for information but often find it is lacking, given the rarity of their child's diagnosis and the uniqueness of their child's symptoms and family's situation.[11] In the hope of improved outcomes, many caregivers conduct their own research, which can be distressing, time-consuming and result in little benefit. Evidence suggests that only ~43% of English-speaking Australians have 'adequate' health literacy.[20] Therefore, many caregivers, if they retrieve information, struggle to decipher whether it is of high-quality or information of relevance for their child. This combination of the 'expert-parent', societal information-seeking behaviour, high social media connectivity of contemporary caregivers and a rapidly evolving genomics field creates the perfect storm for caregivers who are trying to understand their child's complex diagnosis.[11]

Time-poor clinicians may also find it difficult to remain up to date regarding gene-specific knowledge, given the rapidity of gene discovery and publication across both medical and lay resources.[1] This makes it difficult to provide patients and families with the highest-quality information. Communication difficulties can also arise from having a large multidisciplinary team (MDT) that cares for the child, common in rare disease.[21] Limited availability of clinicians with expertise in the specific DEE also results in many families' dissatisfaction with the health system.

In line with the 2020 Australian Government's National Strategic Action Plan for Rare Disease[3] and guided by feedback from families (through both preparatory research[11 12 22] and our consumer reference group), we developed Genetic Epilepsy (GenE) Compass. GenE Compass aims to provide caregivers with greater access to relevant DEE information, promote family-centred care, improve partnerships between researchers and clinicians, and systematically build knowledge and expertise. The previously mentioned research suggests that providing caregivers with understandable, relevant and gene-specific information may help them to better cope with their child's condition.[14]

GenE Compass

We will invite caregivers to submit questions about their child's condition, expected comorbidities, natural history information, support resources, current condition-specific research, or how gene therapies or precision medicine works. We will respond to these questions with individually prepared evidence-based reports. These reports will provide the highest-quality available information that is relevant to their child's diagnosis and be presented in an understandable format.

Objectives

Our mixed-method prepilot–postpilot aims to determine whether GenE Compass is acceptable to caregivers and neurologists and is feasible to deliver. We will also explore the potential impact of GenE Compass on caregivers' quality of life, information searching behaviours, perceptions of their child's illness and activation (ie, willingness and capacity to manage their child's health and care). See table 1 for our logic model.

METHODS AND ANALYSIS

Study design

We developed our protocol to meet the standards outlined in the Standard Protocol Items: Recommendations for Interventional Trials checklist.[23] We will use a single group, predesign–postdesign to achieve our objectives.

Patient and public involvement

Our preparatory research with parents who have a child with a genetic epilepsy led to the innovative design of GenE Compass.[11 12 22] Our protocol has been designed by an MDT which involves a neurologist, a psychologist, a clinical nurse consultant, a genetic counsellor, two clinical geneticists, two behavioural scientists and two implementation scientists. We developed our protocol with these health professionals via online working group meetings over the course of 12 months. We also received input from several other neurologists, paediatricians, clinical nurse consultants, a clinical ethics professional, a medicolegal professional, patient engagement professionals and hospital executives that we identified through our professional networks. We collected this input via email and feedback during two online workshops.

Table 1 GenE Compass logic model.

Inputs	Activities	Outputs	Short-medium term outcomes	Long-term impact
► Funding ► Time ► Experienced medical writer ► Multidisciplinary team with DEE expertise ► Participants	► GenE Compass intervention	► Personalised reports for families and clinicians ► Content on PENNSW website	Caregivers ► Have access to high-quality and relevant information ► Have increased knowledge ► Save time searching for information ► Feel supported	Caregivers ► Feel empowered ► Feel more confident in caretaking ► Experience less distress ► Feel more satisfied with the healthcare system
			Clinicians ► Have increased knowledge ► Save time searching for information	Clinicians ► Feel more confident in caring for their patients

DEE, developmental and epileptic encephalopathy; GenE, Genetic Epilepsy; PENNSW, Paediatric Epilepsy Network NSW.

Four parent consumers contributed toward the development of our questionnaires, and one consumer was involved in the design of GenE Compass and evaluation methods. We collected consumer input via email and online meetings. We have also involved peak bodies such as Genetic Epilepsy Team Australia and the Australian Epilepsy Foundation in the design of our approach to ensure our service is addressing the needs of consumers.

Setting

GenE Compass will be delivered virtually. We will invite caregivers to submit questions online via our purpose-designed form (see online supplemental file A) or via telephone to our information linker, who will complete the online form on behalf of the caregiver. All reports will be delivered via email, and data will be collected online. Should a caregiver not have email or computer access, they will be able to complete all aspects of the study via phone and/or postal mail.

Participants

Caregivers are eligible to participate in GenE Compass if they (1) have a child (<18 years of age at time of study invitation) with a clinically suspected or confirmed diagnosis of DEE; and (2) are new or existing patients at the Sydney Children's Hospitals Network (SCHN). Caregivers whose child transitions to the adult health system during the study will remain eligible. Either one caregiver or both can participate. We will only include caregivers who can speak English. While we acknowledge that culturally and linguistically diverse populations may be the most vulnerable families in this context, further community engagement is needed to ensure the appropriateness of our intervention and study design. Caregivers deemed by one of their clinicians as having significant acute mental health illness, such as currently experiencing suicidal ideation or symptoms of psychosis, will also be ineligible to participate.

Based on a current clinical audit, we anticipate that <10% of the families eligible for our study are likely to become bereaved over the course of data collection. However, it is not possible to predict the life expectancy of children with DEE, as childhood mortality varies significantly between subtypes of DEE. We will confirm appropriateness to send the 3-month follow-up questionnaire (Q2) to participants with their treating team. We will only include bereaved caregivers in our evaluation if they had access to GenE Compass for at least 2 months. In following the recommended length of time in the bereavement literature, we will not contact bereaved caregivers for 3 months following the death of their child.[24–26] At that timepoint, we will call bereaved caregivers to see if they would like to complete a final questionnaire (Q2B) about GenE Compass.

Recruitment

We will obtain a list of eligible families and contact details through study investigators and patient databases including GeneSTART and NeuroCONNECT. GeneSTART is a consented disease agnostic patient database for rare diseases across SCHN. NeuroCONNECT is a registry of patients across the SCHN for neurological disorders.

We will invite eligible families by mailing/emailing an invitation letter and a study postcard with a link to Research Electronic Data Capture (REDCap). This link will include the information sheet and video, e-consent form and Q1, and study contact information. Caregivers can request a paper version of any study documents.

We will recruit families over 4 months. Families will be randomised to one of four recruitment drives via an online randomiser that provides a computer-generated sequence. This stepped approach will ensure we have capacity to conduct follow-up calls, and the information linker can establish a sustainable workflow.

We will follow up caregivers who do not consent to GenE Compass within 2 weeks from the invitation package being mailed out. We will make a maximum of three successful contacts (eg, phone call or email, ie, where contact is directly made with the caregiver). We will also make a

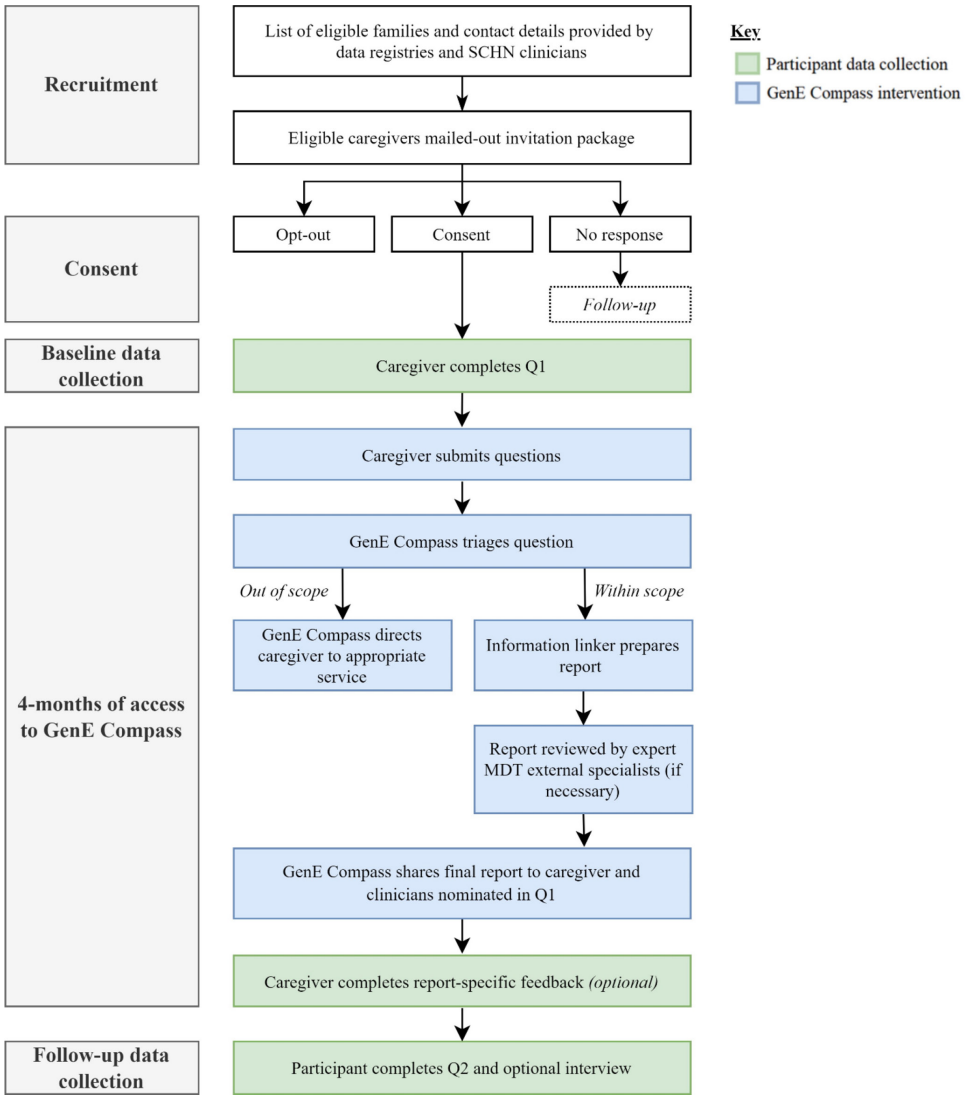

Figure 1 GenE Compass intervention and evaluation process. GenE, Genetic Epilepsy; MDT, multidisciplinary team; SCHN, Sydney Children's Hospitals Network .

maximum of one successful contact (eg, phone call, text message or email) for bereaved caregivers who express an interest in completing Q2B but have not yet done so. Participants will be informed about the voluntary nature of the study and the option to opt out at any point during any follow-up calls.

GenE Compass

Intervention design

Figure 1 outlines the key stages of our GenE Compass pilot intervention, alongside the evaluation data collection points. The key stages of GenE Compass intervention include the following:

1. *Caregiver submits a question.* Following consent and completion of Q1, caregivers are invited to submit questions via an online form (REDCap) or a phone call. Questions can be submitted independently or in partnership with a healthcare professional. This was to empower caregivers and to minimise burden on healthcare professionals.
2. *GenE Compass triages question.* Once we receive a question, our information linker and expert MDT (consisting of a paediatric epileptologist, clinical geneticists, a psychiatrist, a clinical nurse consultant, an epilepsy educator and a genetic counsellor) will triage questions as being (1) within scope or (2) outside of the scope of GenE Compass. GenE Compass has been designed to complement, not replace clinical care. Thus, questions regarding specific management advice (eg, advice on choice of one therapy over another) for an individual patient (or families) are considered outside of scope. Caregivers who submit a question regarding specific management will be prompted to discuss further with their child's clinicians. Submitted questions that can be best responded to by a reputable alternative service/organisation (eg, local support organisations which provide navigational support for navigating public funding for allied health and disability support) are also considered out of scope. Caregivers will

be redirected to the alternative service/organisation as appropriate.

3. *GenE Compass prepares report for questions that are within scope.* Our information linker will conduct a rapid literature review via relevant search engines (eg, PubMed) and review of online resources (eg, orpha.net). The linker will prepare an initial report and consult with our MDT, and if necessary, external specialists.
4. *Sharing of reports.* Each report will be approved by at least two clinicians from our MDT prior to being emailed to participants (via REDCap). Should participants not have an email, we will confirm the best option for them to receive the reports (eg, via postal mail). The report will also be sent to the primary neurologist, paediatrician and primary physician (general practitioner) that the caregiver is required to nominate during Q1 (irrespective of whether their question is submitted in partnership with a healthcare professional).

The decision to allow for a 3-month access period to GenE Compass was driven by funding limitations and an adequate period to determine how frequently parents wish to submit questions (eg, only at consent or at numerous times over a period).

We anticipate that some of the information that will be used for our reports will not be from peer-reviewed articles (eg, web resources). To inform caregivers of quality of evidence used for their report, we will provide a simple a rating of sources used. Our purpose-designed rating system will group sources into three broad categories, depending on scientific quality. Scientific quality is attributed based on the type of peer-reviewed reference (eg, meta-analysis, systematic review and international guideline), the quartile ranking of the journal it was published in and the type of website (eg, government-run or hospital-endorsed).

Evaluation data collection

Participants will complete Q1 at consent, and Q2 3 months later (see figure 1 for evaluation overview, table 2 for overview of measures and online supplemental file B for Q1 and Q2). We will also invite participants to share report-specific feedback at the time of receiving each report. To reduce burden on caregivers, we will inform caregivers that report-specific feedback is optional. We estimate that both Q1 and Q2 will take 20–30 min to complete, and report-specific feedback will take 2 min. All participants will automatically be emailed a link to Q2 3 months after completing Q1, should they be identified as appropriate to contact at that point in time.

We will also invite caregivers to participate in an optional 30 min interview in Q2 so that we can delve further into their experiences and perceptions of GenE Compass. These interviews will allow us to identify barriers and enablers to using GenE Compass to support future service development. Interviews by a trained psychosocial interviewer will be conducted over the phone or in person, depending on caregivers' preference, and will be audio-recorded then deidentified for analysis purposes.

Neurologists nominated by families in Q1 will be invited to provide feedback about GenE Compass via an online questionnaire. We will email a questionnaire link to neurologists who received at least one report over the study period. We will send a maximum of three email reminders, 1 week between each contact, to clinicians who do not respond. See online supplemental file C for the full questionnaire.

We will capture key demographics of participants who do not participate in our study (eg, age of child and time since diagnosis). We will also document recruitment and retention rates, number and type of questions submitted (and date of submission from consenting), format of question submission, time spent preparing each report (eg, staff time writing reports and time spent in meetings to triage reports), time taken to return the report, the number of questions we receive that are out of scope, and the number of caregivers who report more distress after reading their report and require follow-up.

To evaluate the cost and outcomes of GenE Compass, caregivers will report the number of hours per month searching for information related to their child's diagnosis, treatment, symptoms or care. We will also collect the number of calls to the respective hospitals' epilepsy consultant nurse specialists with questions prior to GenE Compass and subsequently.

Measures

We developed our questionnaires in collaboration with caregivers with a child with a DEE and a multidisciplinary steering committee involving child health researchers, implementation scientists, health economists and clinicians. See table 2 for an overview of measures for caregivers.

Semistructured interviews will involve six purpose-designed questions that allow us to explore the questionnaire findings: (1) reason for signing up to GenE Compass, (2) experiences of submitting questions, (3) thoughts on reports, (4) the process of sending reports to healthcare professionals, (5) impact of GenE Compass and (6) recommendations to improve GenE Compass.

Analyses

We will conduct all statistical analyses using SPSS V.24.0.29 or R. We will classify results as statistically significant when the p value is <0.05 (two-tailed). We will use descriptive statistics to report sociodemographics, child medical characteristics, report-specific feedback, acceptability and feasibility.

We will use regression analyses to analyse any change over time in a participant's quality of life (via the Adult Social Care Outcomes Toolkit–Four-Level

Table 2 Overview of measures collected from caregiver participants

Measure	Items (n) and response options	Psychometrics	Description	Q1	Q2	Q2B	Report feedback
Sociodemographics and child medical characteristics							
Demographics and child medical characteristics	16 items; response descriptors vary	Purpose-designed	Items such as child's date of birth and diagnosis, cultural background and language spoken at home	■			
BRIEF	4 items; 1=always to 5=never (items 1–3), 1=not at all to 5=extremely (item 4)	Validated in adults, with high sensitivity[37 38]	BFRIEF to detect inadequate/marginal health literacy	■			
Acceptability and feasibility of GenE Compass (primary outcome)							
Client Satisfaction Questionnaire[39]—adapted	8 items, 4-point Likert scale (response descriptors vary)	Validated in adults,[39] adapted for this study to be relevant to GenE Compass	Measure commonly used to evaluate satisfaction with healthcare services, items cover satisfaction with a service and perceived improvement in self-efficacy		■	■	
Acceptability of GenE Compass	2 items, open-ended	Purpose-designed	Items regarding why participants felt GenE Compass was beneficial to them (if at all) and how it could be improved (if at all)		■	■	
Distress	1 item; no change, more distressed, less distressed, unsure	Purpose-designed	Item to rate any change in distress after reading the report				■
Satisfaction	1 item, 0=not at all to 5=very	Purpose-designed	Item to rate satisfaction with the report information				■
Perceived understanding	1 item, 0=not at all to 5=very	Purpose-designed	Item to rate perceived understanding of the report information				■
Alternative action	1 item; taken no action, discussed with a health professional, conducted own research, reached out to support groups, contacted family/friends, other	Purpose-designed	Item regarding what participants' perceived they would have done for this specific question if GenE Compass did not exist				■
Potential impact of GenE Compass							
Adult Social Care Outcomes Toolkit–Four-Level Self-Completion Questionnaire for Carers	7 items, 4-point Likert scale (response descriptors vary)	Validated in adult carers[40 41]	Measure of aspects of a carer's quality of life that are relevant to, and the focus of, social care interventions	■			
Brief Illness Perceptions Questionnaire[42]	8 items, 11-point Likert scale (response descriptors vary)	Validated in numerous populations[43]	Measure of perceived view of the illness and previously used in caregivers who have a child with a neurological condition[44]	■			
Patient Activation Measure–Short Form[45]—adapted	13 items; 1=disagree strongly to 4=agree strongly, with N/A option	Validated in adults patients,[45] including those with neurological conditions[46]; adapted for this study to be completed by a caregiver	Measure of patient knowledge, skill and confidence for self-management; using this measure, we found that better patient activation appears to be linked to better health outcomes and healthcare satisfaction, and improved self-management.[47 48]	■			
Information searching preferences and experiences	3 items; response descriptors vary	Purpose-designed	Items regarding time spent searching for information over the past month, where information is sourced from and any barriers to seeking information	■			

Note: Q1, baseline; Q2, 3-month follow-up questionnaire; Q2B, bereaved caregivers' follow-up questionnaire; report feedback, report-specific feedback.
BRIEF, Brief Health Literacy Screening Tool; GenE, Genetic Epilepsy; N/A, not applicable.

Self-Completion Questionnaire for Carers (ASCOT Carer–SCT4)), illness perceptions (via the Brief Illness Perceptions Questionnaire (BRIEF IPQ)) and activation (via the Patient Activation Measure (PAM)–Short Form). We will include health literacy and the number of questions submitted to GenE Compass as predictors to this model. We anticipate these variables to be correlated but still have independent effects. However, if they are highly correlated (0.9 or greater), we will include only health literacy as the predictor.

We are collecting data primarily through REDCap, which will limit missing data in Q1. If necessary, we will conduct Little's Missing Completely at Random (MCAR) test to determine whether data are missing completely, and apply multiple imputation using chained equations for missing data on the ASCOT Carer–SCT4, BRIEF IPQ or PAM–Short Form.

We will use NVivo V.12 (QSR International) to conduct a qualitative analysis of the questions submitted to GenE Compass. To increase the depth of findings, we will use an explanatory sequential mixed-method design by using the qualitative data (in interviews) to elaborate on the quantitative (acceptability and feasibility) findings.[27] We

will code transcriptions line by line using an inductive coding approach.[28]

Sample size

The target sample size for this project is 72 participants at baseline, which will allow for an estimated final sample size of at least 50 participants. This assumes a response rate of 20% and an attrition rate of 30%. There is little research in this population to determine our attrition rate. As such, we have based it on experience of the researchers and clinical expertise.

With no research available that specifies minimally important differences for the ASCOT Carer-SCT4, Brief IPQ or the PAM–Short Form, we based our sample size calculation off the research conducted with the Short-Form Six-Dimension (SF-6D) andEuropean Quality of Life-Five Dimension (EQ-5D) preference-based measures of health.[29] We assumed a correlation of 0.6 between repeated ASCOT Carer-SCT4 measures and an SD of 0.3. For a final sample size of n=50, there would be greater than 80% power to detect a change of 0.2.

ETHICS AND DISSEMINATION

Data management

All questionnaire data, questions and feedback will be collected through the secure UNSW Sydney REDCap server. REDCap is a widely used electronic data capture platform designed to replace paper-based questionnaires and spreadsheet-based data capture systems. Only key research staff will have access to the GenE Compass database. Should a participant request to complete hard-copy questionnaires and/or feedback, hard-copy data will be entered into REDCap by the research team. All hard-copy documents will be held securely at UNSW Sydney in a locked filing cabinet accessible by the study team. Electronic databases and information will be stored on the secure UNSW OneDrive accounts which are only accessible by the research team. E-consent forms will be securely stored through the REDCap 'Auto-Archiver+e-Consent Framework' File Repository.

Any identifiable information that is collected about participants in relation to this study will remain confidential and will be disclosed only with participants' permission or except as required by law. Data for each participant will be labelled with a unique identification number. However, data will be reidentifiable if necessary. All information (hard copies and electronic copies) will be confidentially disposed of 15 years from publication. Paper-based documents will be shredded, and all electronic files will be deleted at the specified time.

Ethics

Our study has been approved by the SCHN Human Research Ethics Committee (2021/ETH11277). We will see approval for any important protocol modifications to this committee. This study is listed on the Australian New Zealand Clinical Trials Registry and has undergone rigorous multidisciplinary peer review. We will submit any amendments to our protocol as necessary. Study progress will be submitted to the SCHN HREC as required. As per the National Statement on Ethical Conduct in Human Research,[30] participants will be informed of the voluntary nature of the study and will be able to revoke their consent to participation at any time without needing to provide a reason.

Safety monitoring

We will not require a data monitoring committee as the proposed study poses minimal risk to participants. However, the population we are working with is vulnerable, and there is potential that the information provided by GenE Compass may cause some distress. Our research team will be automatically notified via a REDCap alert should a participant (1) rate increased distress after reading the report and also (2) indicate that they would like to be contacted by the GenE Compass team. We will contact these participants to further assess and instigate appropriate support services. If we determine the participant to be in imminent risk (eg, suicidal intent), we will contact 000. Any adverse events that occur after informed consent is signed, such as increased distress following a report being read, will be recorded in an adverse events log. If distress is identified during an interview, the interview will cease, and we will provide appropriate support services.

We will include a medicolegal disclaimer in all reports, approved by the SCHN medicolegal team, that specifies (1) the report does not act as a substitute for clinical examination and is not intended to constitute medical advice, (2) consultation with relevant healthcare professionals for any clinical questions, (3) attending the local emergency department in an emergency and (4) our recommendation to discuss this report with relevant healthcare professionals. In all participant contacts, we will also provide details for support networks (eg, Lifeline) and advise them to contact 000 if they are in immediate danger.

Given that our trial is of a short duration with known minimal risks, we will not require a data monitoring committee. We anticipate one interim analysis and one final analysis. The trial will not be stopped in case of futility, unless the project team has serious concerns about participant safety.

Study duration

We will commence study recruitment in January 2022. We will close study recruitment when at least 50 participants have completed both Q1 and Q2, or when funding ceases at the end of 2022 (whichever occurs first).

Dissemination of research findings

We will publish the results of this study in a peer-reviewed journal (with authorship defined by the International Committee of Medical Journal Editors [ICMJE] criteria)

and present our findings at relevant scientific conferences and professional meetings. We will send a lay summary to all participants at study close, and share findings with relevant advocacy groups, clinicians and health services. To ensure confidentiality of participants, given the nature of our sample, the sharing of deidentified data will be considered on request.

DISCUSSION

GenE Compass is an innovative, new model of information provision for families of children with a DEE, a complex and severe group of rare genetic conditions. To our knowledge, it will be the world's first evidence-based intervention to systematically address the information needs of caregivers with a child who has a suspected or confirmed diagnosis of DEE. There are few interventions that use a similar tailored information service approach. Of those that exist, they are most commonly to support primary care,[31–35] but none to our knowledge in a rare disease cohort. We partnered with families to codesign our approach and in direct response to their expressed needs and preferences.[11 12] GenE Compass has the potential to improve caregivers' quality of life and well-being, improve caregivers' self-efficacy and confidence in managing their child's condition, and enhance skills and competences of healthcare professionals—all key targets of rare disease plans and policies, such as the Australian National Strategic Action Plan for Rare Disease.[3]

With the patient and caregiver at the core of GenE Compass, it is integral to develop an intervention that can be embedded within the healthcare system. As such, we have engaged front-line care providers (eg, paediatricians and neurologists), health services (eg, hospital executives and patient engagement staff) and patient advocacy groups. What has resulted is a high-quality intervention design that has a high likelihood of integration into statewide and national health services.

To ensure sustainability of GenE Compass, we will use submitted questions and prepared reports to guide the development of resources for families and clinicians. These resources will be uploaded onto the PENNSW website, which will be freely accessible to families and clinicians globally.[36] This pilot evaluation will result in a 'living information resource', tailored to the most frequently asked questions of caregivers of children with genetic DEE, but available free to access to families and clinicians internationally.

Following our pilot, we will revise GenE Compass as appropriate before evaluating the efficacy and implementation of the intervention at a national level. We will also adapt our innovative model of information provision to a broader range of rare neurogenetic diseases, the incidence of which has increased exponentially with advances in genomic technology. GenE Compass therefore has the potential to be relevant to the estimated 2 million people living with rare disease across Australia.[3]

A strength of GenE Compass and our evaluation is in its innovation, but this study is not without limitations. We will investigate feasibility of GenE Compass, but it is likely a resource-intensive intervention requiring substantial funding. This introduces challenges to scalability and sustainability. We will explore cost of GenE Compass (eg, time taken to prepare reports) and acceptability to better understand what aspects of GenE Compass are most impactful to families. We will take these learnings to determine which aspects of GenE Compass may be possible to scale up. A second limitation is the exclusion of the potentially most vulnerable groups—culturally and linguistically diverse families and those who are experiencing acute distress. By limiting recruitment to parents who speak English and who are not experiencing acute psychological distress, there is a potential that we are excluding parents who may be most vulnerable and in need of this service. A further limitation is that the success of this evaluation will largely depend on the recruitment and submission of questions to GenE Compass—it is unclear on the 'dose' that is required to have an impact on participants, or whether 3 months of access provides enough time to reap the potential benefits. Lastly, we recruited from only two hospitals in Sydney, Australia, although this is appropriate for the pilot nature of this study.

Our learnings from this study will provide insight into how we can best provide families with answers to their questions, including the cost–benefit to such an innovative model of care. In addition, our learnings will inform what information families want to know, which can adapted to address the information needs across a broader range of rare diseases.

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Contributors EGR, SMN, RM, FLM, EB, RS, AB and EEP developed the study concept and initiated the project intervention. EGR, LK, SB, IG, EEP and AB contributed to the design of the evaluation. EGR, NG and EEP led the development of an in-depth protocol. EGR and EEP were responsible for the writing this article.

LK, SB, IG, KP, NG, SMN, RM, FLM, EB, RS and AB were responsible for reviewing the manuscript and providing feedback. EGR, NG and EEP implemented the protocol and oversaw the data collection. All authors read and approved the final manuscript.

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