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Families' experiences with pediatric immunology genomic testing: A cross-sectional survey

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Abstract

Genomic testing enables accurate diagnosis and tailored management of paediatric inborn errors of immunity (IEI). However, limited understanding of families' experiences and needs hinders the development of responsive models of care. This study aimed to assess families' experiences with IEI genomic testing and psychosocial wellbeing. Eligible participants included carers of children who underwent IEI genomic testing at <18 years between 2017-2025 in Queensland, Australia, and their adolescent/young adult (AYA) children (≥12 years). Cross-sectional surveys, using validated scales and purposively-designed items, captured participants' experiences with IEI genomic testing, psychosocial wellbeing, and support needs. Cohort characteristics (carers n=42, AYAs n=10) were descriptively summarized and factors associated with carer psychological distress were evaluated using multivariable linear regression. High satisfaction with genomic testing was reported by carers (M:8.5/10;±2.3) and AYAs (M:8.0/10;±1.6) and carers assigned high personal utility to the results (M:4.7/7;±1.1). Over half of carers (n=24/42) reported elevated psychological distress, with significantly higher levels among carers whose child received a positive genetic result ($p<0.05$). Around half of AYAs (self- or proxy-reported) experienced difficulties across health-related quality-of-life domains and ≥20% of carers or AYAs reported clinically significant levels of anxiety, depression, anger and/or need for help. Participants expressed concerns about future reproductive planning (carers: n=23/42;54%, AYAs: n=6/10;60%) and transitioning to adult healthcare (carers: n=9/42;21%, AYAs: n=4/10;40%) and reported a need for condition-specific information (carers: n=9/42;21%; AYAs n=4/10;40%). These findings demonstrate high acceptability towards IEI genomic testing among families while highlighting the importance of tailored psychosocial and informational support.

Key words: Inborn errors of immunity, primary immunodeficiencies, genomic, psychosocial, pediatrics, immunology

Introduction

Inborn errors of immunity (IEI) are an expanding group of genetic conditions that impair the development and/or function of the immune system.[1] These conditions present with a wide range of clinical manifestations, including increased susceptibility to infections, allergies, autoinflammation, autoimmunity, and malignancy.[1] IEI are estimated to affect between 1:1,000 and 1:5,000 individuals, with most of these conditions presenting during infancy and early childhood.[2,3] Over 555 monogenic IEI have been described to date, a marked increase from the 30 conditions initially reported in 1995.[1] The growing number of recognized IEI is largely attributed to advances in genomic technology, which have enabled the identification of novel genetic causes.[4] With over 500 causative IEI genes described to date, genomic testing has become key to facilitating accurate diagnoses and tailored management, including curative therapies for specific conditions.[1]

Living with a rare disease is associated with significant psychosocial challenges for paediatric patients and their families, which often contribute to high levels of emotional difficulties and reduced health-related quality of life (HRQoL).[5,6] While IEI patients and their families often experience similar challenges, they also face distinct concerns arising from complex and/or ongoing treatment regimens and side effects, recurrent infections, physical sequelae of immune dysfunction, and frequent hospitalizations.[7] Among parents of children with an IEI, such factors contribute to heightened anxiety, significant caregiver burden, and reduced HRQoL,[8,9] particularly among those whose child is undergoing intensive treatment.[10] Similarly, pediatric IEI patients report impacts on their physical and social activities and associated fatigue, anxiety, depression, and reduced HRQoL.[11,12] However, these outcomes are primarily drawn from studies that focus on severe IEI and often do not capture paediatric patients self-reported experiences.[9]

Given the clinical utility of genomic testing for pediatric IEI, new models of care are increasingly being developed to promote its optimal integration into routine healthcare.[13-15] Such models of care have been shown to improve genomic testing practices,[15,16] and to be acceptable among parents of children with an IEI.[9] Nonetheless, further exploration of families' experiences living with an IEI and genomic testing is needed to ensure new models of care are equipped to deliver patient-centred care and address families' needs. Thus, this study aimed to assess IEI patients and families: i) prior experiences with the clinical diagnostic and genomic testing process, ii) psychosocial wellbeing, and iii) support needs. This study tested the

hypothesis that psychological distress was higher among carers whose i) child received a positive genomic result, and ii) child's condition had a greater impact on their daily activities and/or educational functioning.

Materials and Methods

Ethics Approval

This study was approved by the Queensland Children's Hospital (QCH) Human Research Ethics Committee (HREC) (HREC/24/CHQ/111751) and the University of Queensland (UQ) HREC (2025/HE001943).

Study Design and Setting

This study is part of the wider BRIDGE (**B**uilding **R**esources for **I**mmunology **D**iagnostic **GE**nomics) project,[9,15-17] which aims to optimize pediatric IEI genomic testing practices by considering the needs of patients, families, healthcare providers, and health services. This project is conducted within the context of the Queensland Pediatric Immunology and Allergy Service (QPIAS), QCH, Australia. A genomic model of care was established within QPIAS in 2021 to support the delivery of genomic testing within routine healthcare.[15] Key features of this model of care, including a genetic counsellor embedded within the service, funding for a clinical immunologist to specialize in genomics, and monthly IEI multidisciplinary team meetings.

This project was designed and conducted alongside three consumer representatives (authors L.G., S.V., and J.P.). Specifically, this study employed a quantitative-dominant mixed-methods approach, utilising cross-sectional surveys to capture the experiences of carers of children who underwent IEI genomic testing and their adolescent/young adult (AYA) children. The term 'carer' is used throughout this study rather than parent, to be inclusive of other family members who may care for the child.

Participant Eligibility

Carers were eligible to participate if their child underwent IEI genomic testing between 2017 and 2025 and was aged 18 years or younger at the time of testing. Eligible families were identified via the local IEI genomic database,[16] and reviewed by the clinical team to confirm eligibility (authors P.M., A.S., M.M., A.P., J. E.P., and T.Y.). Exclusion criteria were developed in consultation with the study consumers (authors L.G., S.V., and J.P.). Specifically, carers were excluded if their child was deceased or was approaching end of life, was undergoing a

hematopoietic stem cell transplantation, had received a clinical or molecular diagnosis in the past two months, or was noted to have significant psychosocial concerns. While the perspectives of these excluded carers are important, it was determined that such factors may lead to heightened distress and that survey invitation may place undue burden on families.

Only AYAs whose carers met the study eligibility criteria and were aged twelve years or older were eligible to participate. This age restriction was selected to ensure AYAs had the necessary reading skills and emotional maturity to complete the survey.

Participant Recruitment

Recruitment occurred August 2025 and January 2026 when eligible carers received study invitations via email, along with the participation information sheet and a link to the carer survey. Carers of eligible AYAs were also sent a link to the AYA participant information sheet and survey, which carers could share with their child at their own discretion.

We sent up to three study reminders to carers via email or follow-up phone call. At this point a lack of response was deemed non-participation. Survey data was collected via the Qualtrics system administered by the University of Queensland.[18]

Carer and AYA participants provided written online consent prior to completing the survey, and AYAs were required to indicate if their carer assisted them in completing the survey. Interpreters were offered to assist non-English-speaking participants.

Survey Measures

Surveys were developed by a multidisciplinary team comprising researchers and clinicians with expertise in genetic counselling, psychology, and pediatric immunology (**Table S1**), and piloted with study consumers (L.G., S.V., and J.P.). The AYA survey included fewer measures to reduce participation burden.

We assessed carers and AYAs' experiences with the clinical diagnostic and genomic testing process using purposively designed items based on previous literature related to other paediatric conditions.[19-21] Participants who did not recall the genomic testing process were not asked about their related distress or satisfaction. The carers survey also captured perceived utility of genomic testing via the Parental Personal Utility Scale (PrU).[22] Psychosocial wellbeing for carers and AYAs was measured using Emotion Thermometers Tool,[23] and the respective EQ-ED-3L parent-proxy and EQ-5D-Y-3L youth measures.[24] The carers survey also assessed general distress via the Kessler Psychological Distress Scale (K10).[25] Finally,

purposively-designed items captured carers' and AYAs' concerns about their/their child's condition and genomic results, as well as their support needs. The carers survey also assessed barriers to accessing support. Both surveys included several open-ended fields to allow participants to elaborate on their answers.

Data analysis

We analyzed quantitative data with the Statistical Package for Social Sciences (SPSS) (version 39).[26] Basic descriptive statistics were used to describe the cohort, including socio-demographics and clinical data. Geographical remoteness was assessed using the Australian Statistical Geography Standard (ASGS),[27] and socio-economic status was determined using the Socio-Economic Indexes for Areas (SEIFA),[28] both calculated via postcode data according to the Australian Bureau of Statistics. Standardized measures were scored according to guidelines (**Table S1**). Continuous variables were described using the mean (M) and standard deviation (±), and categorical variables were summarized as frequencies and proportions (%). Survey responses were included if at least 60% was completed. Where answers were missing due to skipping of questions, denominators included respondents only. For carers who missed one K10 item, the missing value was imputed using the mean substitution method.[29]

Clinical and genomic data (i.e., indication for testing, instances of genomic testing) were extracted from the local IEI genomic database for carer's child with an IEI.[16] In instances where a carer had more than one child with an IEI who had undergone genomic testing, only the proband's clinical and genomic data was reported (i.e., initial child to undergo genomic testing). Additionally, for families in which more than one carer completed the survey, the related proband's clinical and genomic data was reported only once.

We used linear regression models to examine associations between carers' distress (assessed via K10 total scores[25]) and socio-demographic and clinical variables. Categorical variables were included using indicator (dummy) variables, with one category specified as the reference group. Variables with $p<0.20$ in unadjusted analyses were considered for inclusion in the multivariable model. A backward stepwise approach was applied, with variables sequentially removed based on Wald tests for overall significance. The final model was adjusted for distance from residential address to QCH, number of days missed from school in the past year, indication for genomic testing, and molecular diagnosis. Results are presented as regression coefficients (β) with 95% confidence intervals (CIs). Regression analyses were conducted using STATA (version 19)[30] with $p<0.05$ considered statistically significant. A sample size

calculation of carer participants was conducted *a posteriori,* considering a pragmatic approach to the number of participants included in the study.

Narrative data from open-ended questions were analyzed using qualitative content analysis,[31] via NVIVO (version 15).[32] Illustrative quotes are provided throughout the text to support, clarify, or expand upon the main numerical findings, for which pseudonyms are used.

Results

Participants

Of the 322 patients identified who underwent IEI genomic testing from 2017 to 2025, 145 carers and 66 AYAs were eligible to participate. Following invitation, 15 carers actively opted out due to lack of interest or inappropriate timing. The 42 carers who participated (response rate: 29%) represented 40 unique family units, with two families having two carers each completing the survey. Ten AYAs participated in the study (response rate: 15%), representing nine unique families, with two AYAs from the same family completing the survey. All 10 AYAs and 40 carers completed the entire survey. Four AYAs reported that their carer assisted them in completing the survey. Due to the small number of AYA participants, matched carer-AYA survey data was not included to preserve participant confidentiality. Based on the response rate of carer participants (29%), the estimated margin of error with 95% confidence is low (±7%).

Socio-Demographics Characteristics

Most carers identified as female (n=37;88%), with a mean age of 41.9 years (range: 27-57 years) (**Table 1**). Majority were employed at least part-time (n=29;69%), university educated (n=25;60%), married or in a de facto relationship (n=34;81%), and reported a household income of >$120,000 (n=22;52%). Over half of carers had ≥2 children (n=33;79%), and eight (19%) had >1 child with a chronic health condition. Geographical distance from home to hospital ranged from 1-1696km, although most lived in a major city (n=31;73.8%) and around half resided in an advantaged/most advantaged area (n=20;48%). Most carers identified as White-European (n=33;79%). One-third of carers (33%) reported that their child spent >10 days in hospital in the past year, and 50% reported that their child missed more >10 days of school in the past year. The mean age among AYA respondents was 16.7 years (range: 12-20 years), with most identifying as female (n=6;60%).

Clinical and Genomic Characteristics

On average, children of carers (data reported once per unique family, n=40 children) received a mean of 1.6 genomic tests (range:1-3 tests) and were aged 5.6 years (range:0-16 years) at the time of their initial test (**Table 2**). The type of genomic testing applied within this population has been reported previously.[33] The most common indications for testing were immune dysregulation (n=8;20.0%) and predominantly antibody deficiency (n=7;18%). Eleven children (28%) received a molecular diagnosis (positive result). The most common treatment received was immunomodulation (n=14;35%) and immunoglobulin replacement therapy (n=12;30%).

Diagnosis and Genomic Testing Experience

All but one carer (n=41;98%) and most AYAs (n=8;80%) recalled the genomic testing process. Carers rated receiving their child's clinical diagnosis as the most distressing event (M:6.5/10; ±3.1), followed by waiting for genomic test results (M:5.5/10; ±3.1), receiving such results (M:5.0/10; ±3.1), and deciding to undergo testing (M:3.1/10; ±2.5) (**Figure 1a**). Some carers attributed their distress to the severity of their child's presentation at the time of genomic testing.

"My distress scores were so high as my son had multiple severe infections at the point of [genomic] *testing; I was highly anxious about everything!"* Rebecca (mother), positive genomic result

AYAs reported finding out about their genomic test results (M:4.6/10; ±3.1) as slightly more distressing than living with their health condition (M:3.9/10; ±2.7) (**Figure 1b**). Some AYAs reported that their distress stemmed from a limited understanding of the potential implications of their genomic results.

"I was pretty scared to find out my genetic test results because I didn't know what they were or what they meant." Liam (male AYA), negative genomic result

Regarding the decision to undergo genomic testing, one third of carers reported that their child was involved in the decision-making process, while half (n=21) noted that their child was too young to be involved. Children aged 0 to 7 at the time of initial genomic testing were less likely to be included in the decision-making process (n=4/27;15%) than those aged 8 to 16 years (n=10/15;67%) (**Figure S1**). Among the 27 carers whose child was not involved in the decision-making process, four (10%) reported that they would have liked them to have been

involved, while one-third (n=14;33%) reported being unsure. Of the AYAs, six (60%) noted they were not involved in the decision-making process, with only two indicating that they would have liked to have been.

High satisfaction with the genomic test process was reported by carers (M:8.5/10; ±2.3) and AYAs (M:8.0/10; ±1.6) (**Figure 1**). Both cohorts reported being satisfied with their understanding of the genomic results (carers: M:7.7/10; ±2.5, AYAs: 8.6/10; ±1.5). Furthermore, carers were highly satisfied with having a consistent team who took responsibility for their child's healthcare (M:9.0/10; ±1.6) and the amount of information received about their child's genomic results (M:8.1/10; ±2.3).

In relation to the genomic testing process, over half of carers (n=23;54.8%) recalled meeting with a genetic counsellor. However, half of these carers did not understand the reason for the genetic counselling appointments (n=22;52.4%). Furthermore, some carers noted confusion arising from conflicting information received from different specialties post-result disclosure, typically stemming from differing result interpretations.

"Conflicting information was provided between the genetic counsellor and the immunology registrar, which made the process more difficult." Nicole (mother), negative genomic result

Carers' perceived utility of genomic testing, measured via PrU,[22] was high overall (M:4.7/7; ±1.1) and consistent across all subscales, including child benefits (M:4.6/7; ±1.2), affective parent benefits (M:5.0/7; ±1.3), and parental control (M:4.7/7; ±1.6) (**Figure 2**). Carers reflected on the personal utility of genomic testing in providing diagnostic clarity and clear treatment pathways.

"It [genomic testing] *answered a lot of questions regarding familial history that we otherwise would not have connected the dots."* Jade (mother), positive genomic result

Psychosocial Wellbeing

Across the emotional thermometer tool,[23,34] some carers and AYAs reported clinically significant (≥4) levels of distress (careers: n=4;10%, AYAs: n=1;10%), anxiety (careers: n=12;29%, AYAs: n=2;20%), depression (carers: n=4;10%, AYAs: n=3;30%), anger (carers: n=9;21%, AYAs: n=1;10%), or a need for help (carers: n=6;14%, AYAs: n=2;20%) (**Table S2**).

Carers' distress scores, assessed via K10,[25]were on average consistent with moderate distress (M:18.4/50; ±7.7). Specifically, over half the carers reported elevated distress, with moderate distress reported by 14 (33%), high distress by six (14%), and very high distress by four (10%).

In the multivariable linear regression model (**Table 3**), carers' distress was not found to be associated with distance from residential address to QCH, number of days missed from school in previous year, and clinical indication for genomic testing. However, carers of children who received a molecular diagnosis had, on average, 7.13 points higher psychological distress scores (95% CI: 0.61 to 13.65, p=0.033) compared to carers of children without a molecular diagnosis.

On average, carers rated their child's overall health, as assessed by the EQ-VAS, as 75.6 (out of 100, ±22.3) and AYAs rated their overall health as 71.9 (out of 100, ±8.1), with the index value for carers (M:0.76; ±0.04) similar to AYAs (M:0.77; ±0.08). Around half of carers and AYAs reported at least some difficulties across more than one QoL domain (carers: n=20;48%, AYAs: n=5;50%). Carers most frequently reported difficulty for their child related to feeling, worried, sad, or unhappy (n=20;48%), followed by pain or discomfort (n=16;38%), and difficulties with usual activities (n=15;36%). A few carers also noted that their child has difficulties with self-care (n=7;17%) and mobility issues (n=6;14%) (**Figure 3**). AYAs most commonly reported difficulties with doing their usual activities (n=6;60%), problems with self-care (n=3;30%) and pain or discomfort (n=3;30%). Lastly, two AYAs (20%) noted having mobility issues and feeling worried, sad, or unhappy (20%). Some AYAs further commented on how their condition posed challenges to participating in their usual activities.

"I find it difficult to do university and felt locked out from school when everything went down with my health condition…getting a job because of my immune condition has also been difficult." Archie (male AYA), variant of uncertain significance genomic result

Concerns and Support Needs

Over half of carers (n=24;57%) and 80% of AYAs (n=8) reported having at least one concern about how their condition and genomic results might affect their future. Common concerns among both groups included the potential impact on their future family planning (carers: n=23;54%, AYAs: n=6;60%) and the process of transitioning from pediatric to adult healthcare (carers: n=9;21%, AYAs: n=4;40%) (**Table S3**). Carers also commonly reported concerns about the impact on their child's ability to obtain life insurance (n=16;38%) and AYAs also noted concerns about the possible impact on getting a job (n=4;40%). Several carers further commented on the challenges of transitioning from pediatric to adult care.

"My child's transition to the adult hospital has been horrific and they have absolutely no clue how to deal with her now." Claire (mother), negative genomic result

Around half of carers (n=22;52.4%) and most AYAs (n=8;80%) reported having at least one unmet support need (**Table S3**). Carers most frequently indicated a need for more information about their child's condition and genomic results (n=9;21%) and additional resources to facilitate discussions with their child about their condition and/or genomic results (n=7;17%). Some carers highlighted the lack of family-friendly, condition-specific resources available.

"It's extremely difficult receiving a diagnosis of such a rare condition with no specific support service or information in existence apart from medical journals." Natalie (mother), positive result

Additionally, some carers noted a need for greater financial support (n=5;12%) and access to peer support to connect with other affected families (n=5;12%), specifically noting a desire to connect with others during the initial diagnostic period.

"I wanted to be able to connect with other families who might be able to shed some light on the condition." Natalie (mother), positive genomic result

Some AYAs similarly reported a need for more information about their condition (n=4;40%) and greater social and emotional support (n=2;20%). Several AYAs noted that limited understanding of their condition among peers made it difficult to discuss their support needs.

"I sometimes find it difficult to communicate well with others without support because I can become confused due to pain or past experiences [that] *trigger my anxiety and thinking patterns."* Ruby (female AYA), variant of uncertain significant genomic result

Among carers, the most common barriers to accessing support were having other responsibilities (n=20;48%), feeling that they should be able to their manage concerns themselves (n=18;43%), not knowing where to look or who to ask for help (n=17;41%), and not having access to appropriate resources (n=16;38%) (**Figure S2**).

Discussion

This study provides a novel insight into families' experiences with genomic testing for pediatric IEI, including their psychosocial wellbeing and support needs. Our findings align with previous studies, demonstrating that carers view the decision for their child to undergo IEI genomic testing as straightforward, compared to other medical decisions, and assign high personal utility to genomic results.[35,36] Families were satisfied with having undergone genomic testing as part of routine healthcare, and particularly valued having a consistent healthcare team. However,

families reported reduced psychological wellbeing, requiring additional psychosocial and informational support. Collectively, these findings identify areas where models of care can be adapted to more effectively address families' support needs and optimize the delivery of genomic testing.

Carers and AYAs reported high satisfaction with genomic testing. Although the cohort size did not permit statistical comparisons between responses or groups, our findings are broadly consistent with other studies. In particular, carers reported higher distress in relation to receiving their child's clinical diagnosis and lower distress related to the genomic testing process.[37] Although carers viewed the decision for their child to undergo genomic testing as minimally distressing, higher scores were reported in relation to waiting for and receiving the results.[38] AYAs reported slightly higher distress scores in relation to finding out about their genomic results than living with their health condition, possibly due to the uncertainty of genomic results relative to the known, lived-experience of their condition.[39] Additionally, AYAs' engagement in decision-making related to genomic testing varied by age, with those aged eight years or older more likely to be involved. This finding is consistent with paediatric oncology studies, in which parents and adolescents expressed interest in involving children in such discussions around a similar age.[40] Current guidelines recommend that AYAs are engaged in decision-making related to genomic testing,[41-43] with benefits of including AYAs in discussions about their healthcare including reduced anxiety and increased sense of control.[44] Thus, it is important that models of care consider strategies and resources to engage AYAs in age-appropriate discussions related to genomic testing.

Families within this study reported decreased psychological wellbeing, consistent with the prior IEI literature. Clinically significant levels were reported on each of the five emotional thermometers, with ≥20% of AYAs and/or carers flagging anxiety, depression, anger or need for help, as similarly observed in paediatric oncology.[19] Around half of AYAs and carer-proxies reported at least some difficulties across more than one QoL domain, with nearly half of carers reporting that their child was at least sometimes sad, worried or unhappy, and majority of AYAs noting problems performing their usual activities. Additionally, over half of carers reported moderate or greater psychological distress, mirroring pediatric oncology and rare disease settings.[21,45] In line with our hypothesis, carers of children with a molecular diagnosis had significantly higher distress, likely reflecting the more complex clinical course and the familial and reproductive considerations associated with such results.[46] As heightened distress can impair processing of complex information,[47] careful timing of genomic testing discussions is

essential, and strategies to identify those at risk for heightened distress, such as implementing a patient screening tool, may guide targeted support.[48]

While carers and AYAs were satisfied with having undergone genomic testing, participants also expressed concerns about the future implications of the IEI diagnosis and genomic results. Both groups reported concerns about future reproductive planning, which has similarly been reported in the pediatric oncology setting.[49] Such findings highlight the importance of ongoing genetic counselling to ensure AYAs understand their condition, genomic results, and reproductive options, particularly given that initial diagnostic and genomic testing discussions typically occur at a young age.[50] Efforts are also needed to ensure families understand the role of genetic counselling and receive consistent information. Additionally, transitioning from pediatric to adult healthcare was a common concern among carers and AYAs, which is recognized as a vulnerable period.[51] Increasingly, there are efforts to develop structured and standardized approaches for IEI transition of care. Such approaches recommended early and ongoing discussions about the transition process to support patients' understanding of and engagement in their healthcare, transition timing that is developmentally appropriate, joint consultations with paediatric and adult teams prior to transition, and effective transfer of clinical information.[52,53] It is particularly important that patients' understanding of their genomic results and the associated reproductive implications is supported during the transition process. Furthermore, both groups indicated a need for more information about their condition and genomic results, and carers noted a need for resources to facilitate discussions with their child. Emerging opportunities in digital technology in the field of genetic counseling offer promise in improving access to family-friendly condition-specific information.[54] Finally, carers reported concerns about their child's ability to obtain life insurance. While the Australian federal government has recently introduced legislation banning the use of genomic information in life insurance underwriting, continued efforts are needed to help families understand these changes and address their related concerns.[55] Collectively, these findings identify families psychological and informational support which extend beyond the initial diagnostic period.

These findings should be interpreted in line with the study limitations. For example, the carer cohort primarily consisted of highly educated, female carers of White European ancestry. Given that language, educational, and cultural barriers can affect individuals' experiences, the generalization of our findings is limited accordingly. The sample size was also finite, which limited statistical analyses, particularly among AYA participants. Future studies of larger, longitudinal cohorts should consider quantitatively testing the study observations. Additionally,

purposively-designed items should be validated in a larger cohort. Self-selection bias was also likely present, and the eligibility criteria may have excluded families experiencing heightened distress. Families' experiences may have also varied depending on whether they received testing as part of the model of care and meet with a genetic counsellor during the testing process. Nevertheless, this study has several strengths, including the inclusion of AYAs' self-reported perspectives and families who received genomic testing over an extended period, with a range of genomic results. Additionally, this study is part of a wider evaluation (BRIDGE project),[9,15-17] which allows for triangulation of findings alongside other studies. Overall, our findings demonstrate families are highly satisfied with and accepting of receiving IEI genomic testing within routine healthcare. As genomic testing becomes increasingly integrated into pediatric healthcare, it is imperative that models of care are equipped to adequately address families' psychosocial and informational needs.

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Conflict of Interest Statement

The authors declare no competing conflicts of interest.

Authorship Contributions

E.D. contributed to study conceptualization, data collection and analysis, and writing original draft and editing. L.V-G. and A.C. contributed to data analysis and writing review and editing. T.C., contributed to study conceptualization and project administration. B.M. and K.H. contributed to study conceptualization and writing review and editing. S.B., and D.Z. contributed to writing review and editing. L.G., S.V., and J.P. (study consumers) contributed to study conceptualization and writing review and editing. A.P., M.M., A.S., and J.E.P. contributed to data collection and writing review and editing. A.M. and P.M contributed to study conceptualization, supervision, and writing review and editing. T.Y. contributed to study

conceptualization, funding acquisition, data collection and analysis, and writing review and editing.

Tables

Table 1: *Carers' socio-demographics characteristics*

Carer characteristics (n=42)	
Age, mean (range), years	41.9 (27-57)
Gender, n (%)	
Female	37 (88)
Male	5 (12)
Number of children, n (%)	
One child	9 (21)
Two children	19 (45)
Three children	9 (21)
More than three children	5 (12)
Number of children with chronic health condition, n (%)	
One child	34 (81)
Two children	4 (10)
More than two children	1 (2)
Prefer not to say	3 (7)
Relation to child with an IEI, n (%)	
Mother	36 (86)
Father	5 (12)
Other family member	1 (2)
Education level, n (%)	
High school	5 (12)
Apprenticeship, certificate, or diploma	9 (21)
University degree	25 (60)
Other	3 (7)
Marital status, n (%)	
Currently married or de facto	34 (81)
Separated or divorced or previously de facto	8 (19)
Employment status, n (%)	
Employed full time	20 (48)
Employed part time	9 (21)
Employed casually	3 (7)
Studying full time	1 (2)
Home duties	6 (14)
Retired	1 (2)
Prefer not to say	2 (5)
Household income, n (%)	
Less than $30,000	1 (2)
$30,000 to $59,999	2 (5)
$60,000 to $89,999	4 (10)
$90,000 to $199,999	4 (10)
> $120,000	22 (52)
Prefer not to say	9 (21)
Ethnicity, n (%)	
White European	33 (79)
South-East Asian	2 (5)
Torress Strait Islander	1 (2)

Latin American	1 (2)
Prefer not to say	5 (12)
Geographical classification (ASGS), n (%)	
Major city	31 (74)
Inner regional	7 (17)
Outer regional	4 (10)
Socio-economic status (SEIFA), n (%)	
Most advantaged	9 (21)
Second most advantaged	11 (26)
Middle	15 (36)
Second most disadvantaged	5 (12)
Most disadvantaged	2 (5)
Distance from residence to treating hospital (QCH), mean (range), kilometers	202 (1-1696)
Number of days child missed from school in past year due to their health condition, n (%)	
None	12 (29)
One to five days	2 (5)
Six to ten days	4 (10)
More than ten days	21 (50)
Unsure	3 (7)
Number of days child spent in hospital in past year due to their health condition, n (%)	
None	7 (17)
One to five days	8 (19)
Six to ten days	8 (19)
More than ten days	14 (33)
Unsure	5 (12)

Abbreviations: ASGS; Australian Statistical Geography Standard, IEI; inborn errors of immunity, QCH; Queensland Children's Hospital, SEFIA; Socio-Economic Indexes for Areas

Table 2: *Clinical and Genomic Characteristics*

Clinical characteristic (n=40 probands)	
Child's age at initial genomic test, mean (range), years	5.58 (0-16)
Number of genomic tests child received, mean (range)	1.58 (1-3)
Child's genomic diagnosis status, n (%)	
Yes	11 (28)
No	29 (73)
Child's treatment, n (%)	
Immunomodulation	14 (35)
Immunoglobulin Replacement Therapy (IRT)	12 (30)
Hematopoietic Stem Cell Transplantation (HSCT)	7 (18)
Antibiotics	3 (8)
No immunology treatment	4 (10)
Child's indication for testing, n (%)	
Immune dysregulation	8 (20)
Predominantly antibody deficiency	7 (18)
Autoinflammation	5 (13)
Atypical infection	5 (13)
Very Early Onset Inflammatory Bowel Disease (VEO-IBD)	5 (13)
Combined Immunodeficiency	3 (8)
Severe Combined Immunodeficiency (SCID)	3 (8)
Susceptibility to infections	2 (5)
Autoimmune Lymphoproliferative Syndrome	1 (3)
Interstitial lung disease	1 (3)

Table 3: *Multivariable Linear Regression Analysis*

	n (%)	Carer distress Mean (SD±)[a]	Unadjusted β (95%CI)	*p*	Multivariable β (95%CI)	*p*
Distance from residential address to Queensland Children's Hospital				0.172		0.232
<50 kilometers	23 (54.8)	17.45 (±7.78)	1.00		1.00	
>50-200 kilometers	11 (26.2)	22.0 (±7.20)	4.55 (-1.06-10.15)		3.39 (-2.32-9.09)	
>200 kilometers	8 (19.0)	15.86 (±6.96)	-1.60 (-8.19-4.99)		1.01 (-6.29-8.32)	
Number of days missed from school in the previous 12 months[b]				0.475		0.225
None	12 (30.0)	18.17 (±7.21)	1.00		1.00	
One to ten days	6 (15.0)	15.50 (±3.45)	-2.67 (-10.66-5.33)		-4.39 (-11.86-3.08)	
>10 days	19 (47.5)	19.95 (±9.05)	1.78 (-4.12-7.68)		2.16 (-3.52-7.83)	
Indication for genomic testing				0.434		0.309
Predominantly antibody deficiency, susceptibility to infection, atypical infection	13 (32.5)	16.0 (±9.48)	1.00		1.00	
Combined immunodeficiency (CID) and severe combined immunodeficiency (SCID)	6 (15.0)	20.50 (±6.66)	4.50 (-3.18-12.18)		4.41 (-5.44-14.26)	
Immune dysregulation, autoimmune lymphoproliferative syndrome, interstitial lung disease, very early onset inflammatory-bowel disease (VEO-IBD)	15 (37.5)	20.27 (±6.22)	4.27 (-1.63-10.17)		5.17 (-0.51-10.85)	
Autoinflammation	6 (15.0)	17.0 (±7.48)	1.00 (-6.68-8.68)		1.01 (-6.29-8.32)	
Molecular diagnosis				0.061		**0.033**
Yes	11 (27.5)	22.09 (±10.20)	5.06 (-0.25-10.36)		7.13 (0.61-13.65)	
No*	29 (72.5)	17.03 (±6.10)	1.00		1.00	

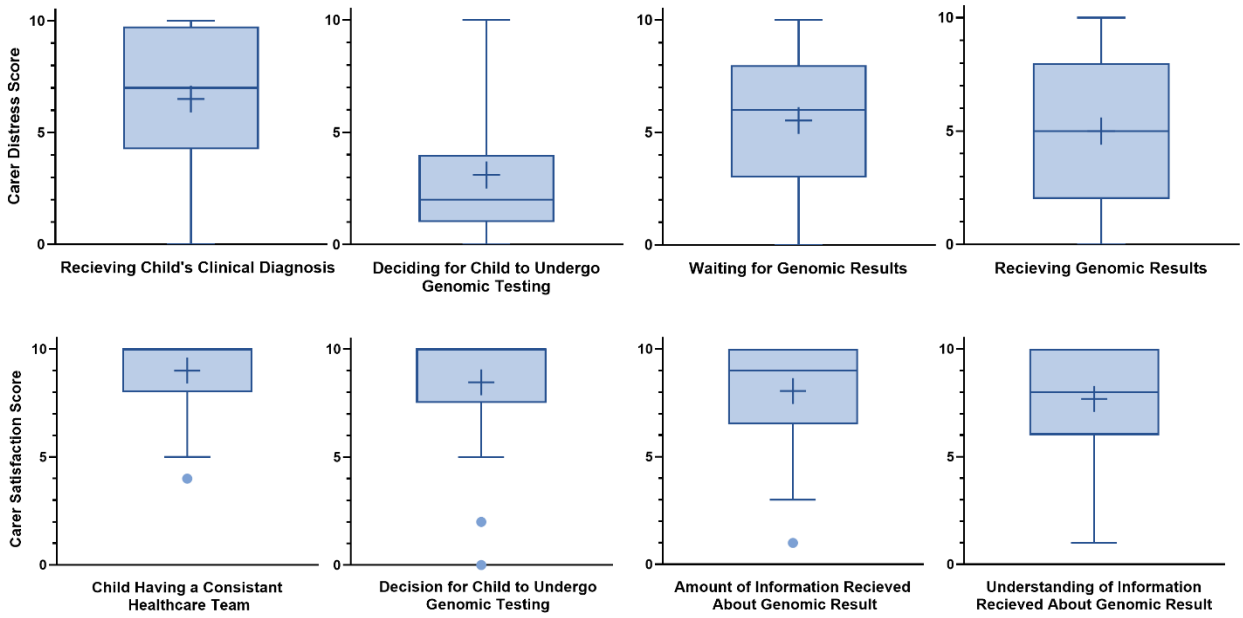
P-values are from the Wald test to assess the overall effect of the variable.

[a]Measured using the Kessler Psychological Distress Scale 10 (K10) total score. Missing responses were not considered (n=2).

[b]Unsure responses for number of days missed from school were not included (n=3).

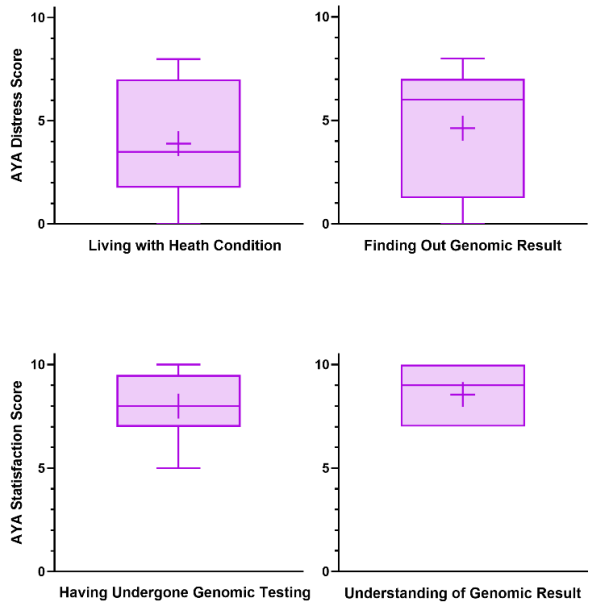
Figures

Figure 1a: Carers' Distress and Satisfaction with Clinical Diagnostic and Genomic Testing Process (n=42)



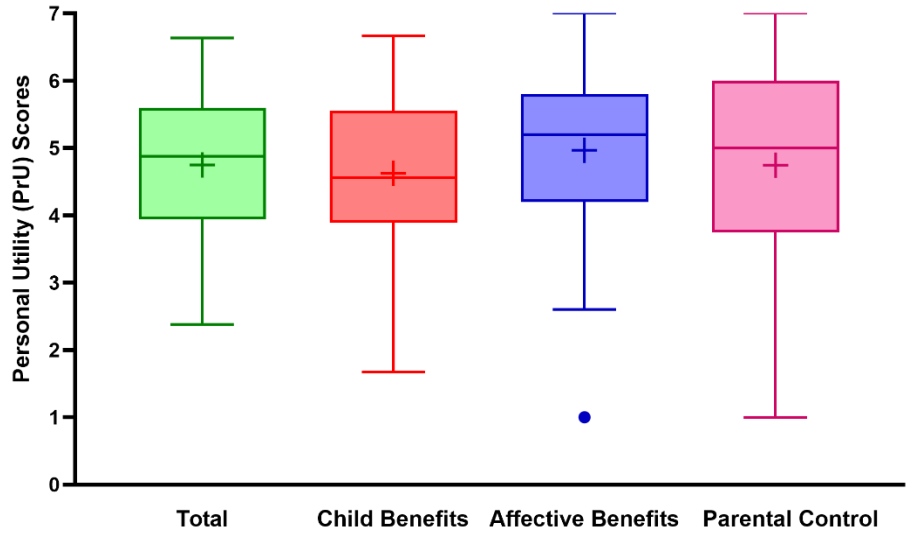
Scale range 0=not applicable to 10=extremely distressing/satisfied. Box plot includes median (+), interquartile range (IQR; 25th-75th percentiles) demonstrated by box edges, minimum and maximum values indicated by whiskers, and points outside the whiskers represent outliers.

Figure 1b: AYAs' Distress and Satisfaction with Clinical Diagnostic and Genomic Testing Process (n=10)



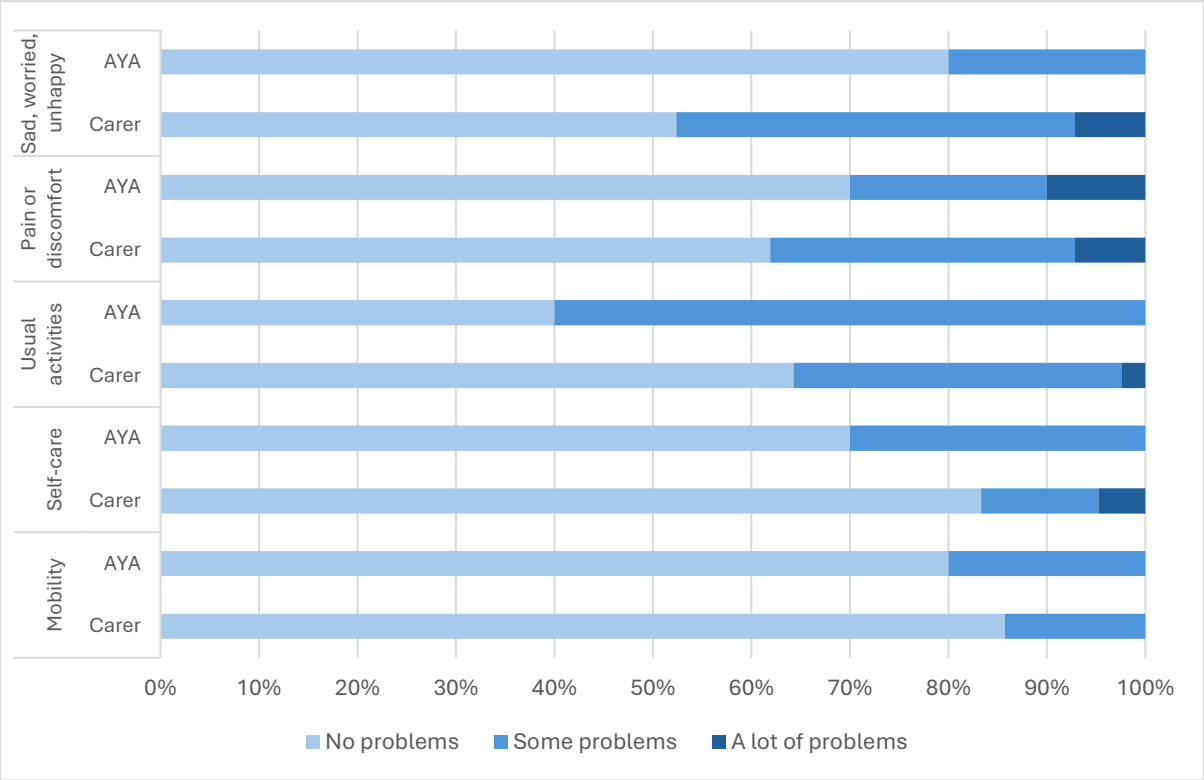
Scale range 0=not applicable to 10=extremely distressing/satisfied. Box plot includes median (+), interquartile range (IQR; 25th-75th percentiles) demonstrated by box edges, minimum and maximum values indicated by whiskers, and points outside the whiskers represent outliers.

Figure 2: *Carers perceived personal utility of their child's genomic testing (n=42)*



Score range 1=not at all useful to 7=extremely useful. Measured via Parental Personal Utility Scale (PrU). Box plot includes median (+), interquartile range (IQR; 25th-75th percentiles) demonstrated by box edges, minimum and maximum values indicated by whiskers, and points outside the whiskers represent outliers.

Figure 3: Health-related quality of life: self-reported by AYAs (n=10) or proxy-reported by carers for their child (n=42)



Measured via the EQ-ED-3L parent-proxy and EQ-5D-Y-3L youth measures respectively. Matched data not shown due to small sample size. Abbreviations: AYA; adolescents and young adults

Supplementary material

Table S1: Description of survey measures and items

Construct	Measure	Respondent	Item/description of measure and scoring	Response options	Reliability/ validity
Socio-demographics	Purposively designed items	Carer	Age, gender, postcode, education level, marital status, employment status, income, ethnicity, number of children, number of children with chronic health condition, relationship to child, and number of days child missed from school and spent in hospital in previous 12 months due to their health condition	Multi-choice	Informed by previous studies
		AYA	Gender and age		
Diagnosis and genomic testing experiences	Purposively designed items	Carer	Recall of genomic testing Distress and satisfaction levels related to aspects of the diagnostic and genomic testing process (e.g., receiving clinical diagnosis, deciding to undergo genomic testing)	Recall of genomic testing: Yes, No Distress and satisfaction scales: 0=I don't remember/not applicable to 10=extremely distressing/satisfied Open-text field: experience with genomic testing	Informed by previous studies[19]
		AYA			
	Parental Personal Utility Scale (PrU)[22]	Carer	Three subscales, including child benefits, affective parent benefits, and parental control. Each subscale is scored as the mean of its constituent items, and an overall total score is derived as a mean of all items. All scores range from 1-7, with higher scores indicating greater personal utility	16-item scale 1=not at all useful to 7=extremely useful	Validated measure[22]
Psychosocial wellbeing	Emotion thermometers tool	Carer	Five domains, including distress, anxiety, depression, anger, and need for help. Scores ≥4 across any domain considered clinically significant	10-point visual thermometer score 0=none to 10=extreme	Validated measure[23]
		AYA			
	Kessler psychological distress scale (K10)	Carer	Total score range 10-50 with higher scores indicating greater distress. Scores categorized as low (10-15), moderate (16-21), high (22-29), and very high (30-50)	10-item scale 1=none of the time to 5 = all the time	Validated measure[25]
	EQ-5D-3L parent proxy version	Carer	Five domains, including mobility, self-care, doing usual activities, pain or	Five domains scale 1=no problems,	Validated measures[24]

	EQ-5D-Y-3L youth version	AYA	discomfort, and feeling worried, sad or unhappy, which participants rate the extent to which their child's /they experience problems Visual analogue scale (EQ-VAS) on which participants rate their child's/their overall HRQoL respectively	2=some problems, 3=a lot of problems VAS scale 0=worst imaginable health to 100=best imaginable health Index value score 1=perfect health to 0=death, analyzed per the Australian sample value set[56]	
Concerns and support needs	Purposively designed items	Carer	List of potential support needs and concerns related to their child's health condition and/or genomic results List of barriers to accessing support and potential unmet support needs	List of concerns and support needs: Yes, No Barriers to accessing support: 14-item scale 1=never to 4=always Open-text field: Support needs	Informed by previous studies[20,21]
		AYA	List of potential support needs and concerns related to their health condition and/or genomic results	List of concerns and support needs: Yes, No Open-text field: Support needs	

Abbreviations: AYA; adolescent and young adult, HRQoL; health-related quality of life

Table S2: Psychosocial wellbeing

	Carer (n=42)	AYA (n=10)
Emotion Thermometer Tool score ≥4*	**n (%)**	**n (%)**
Distress	4 (10)	1 (10)
Anxiety	12 (29)	2 (20)
Depression	4 (10)	3 (30)
Anger	9 (21)	1 (10)
Needing help	6 (14)	2 (20)
Kessler psychological distress scale (K10)		
Low (score 10-15)	16 (38)	NA
Moderate (score 16-21)	14 (33)	NA
High (score 22-29)	6 (14)	NA
Very high (Score 30-50)	4 (10)	NA
Nil response	2 (5)	NA
EQ-ED-3L parent-proxy and EQ-5D-Y-3L youth respectively		
At least some difficulty across 1 QoL domain	5 (12)	2 (20)
At least some difficulty across 2 QoL domains	8 (19)	3 (30)
At least some difficulty across 3 QoL domains	6 (14)	1 (10)
At least some difficulty across 4 QoL domains	5 (12)	0 (0)
At least some difficulty across 5 QoL domains	1 (2)	1 (10)

Abbreviations: AYA; adolescent and young adult

*score ranges from 0=none to 10=extreme

Table S3: Concerns and support needs

	Carer (n=42)	AYA (n=10)
Concerns about how child's health condition and/or genomic results might affect:	**n (%)**	**n (%)**
Ability to obtain life insurance		
Yes	16 (38)	1 (10)
No	26 (62)	9 (90)
Ability to get a job		
Yes	8 (19)	4 (40)
No	34 (81)	6 (60)
Transition from pediatric to adult service		
Yes	9 (21)	4 (40)
No	33 (79)	6 (60)
Family planning		
Yes	23 (55)	6 (60)
No	19 (45)	4 (40)
Support that would be most helpful:		
Resources to help you talk to your child about their health condition and/or genomic test results		
Yes	7 (17)	NA
No	35 (83)	NA
Financial support		
Yes	5 (12)	NA
No	37 (88)	NA
More information about your child's health condition and/or genomic test results		
Yes	9 (21)	4 (40)
No	33 (79)	6 (60)
Practical support		
Yes	3 (7)	NA
No	39 (93)	NA
School support		
Yes	3 (7)	NA
No	39 (93)	NA
Peer and/or community support		
Yes	5 (12)	1 (10)
No	37 (88)	9 (90)
Emotional support		
Yes	3 (7)	2 (20)
No	39 (93)	8 (80)

Abbreviations: AYA; adolescent and young adult

Figure S1: Carers child's involvement in the decision to undergo genomic testing (n=42)

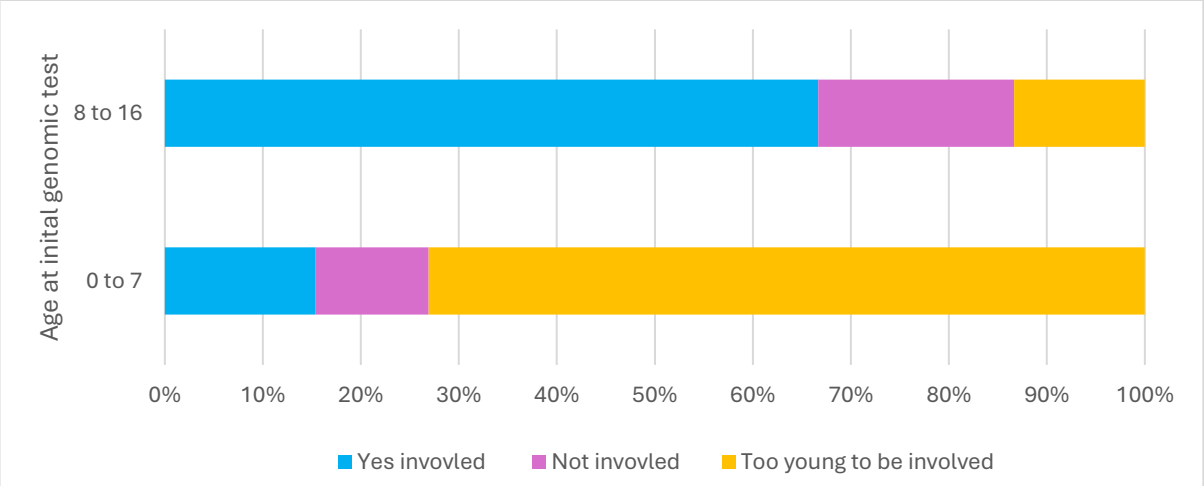
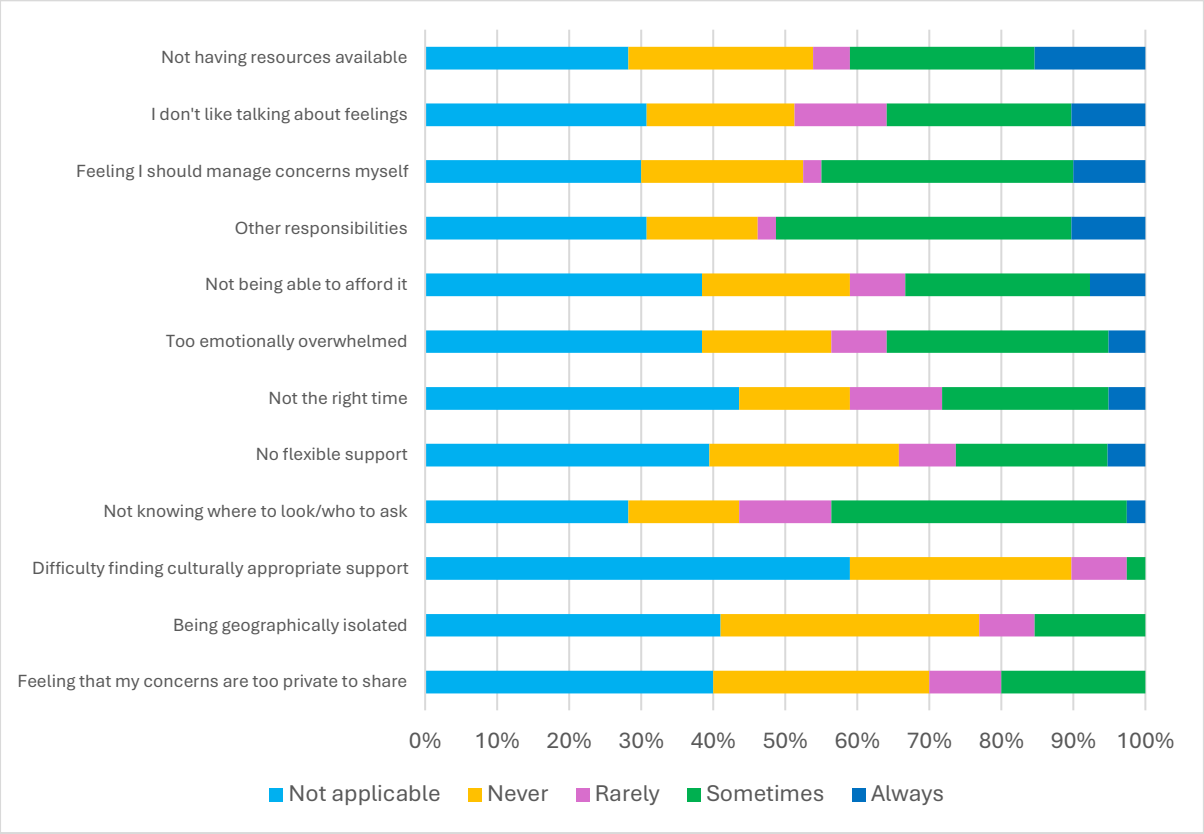


Figure S2: Careers' perceived barriers to accessing support (n=42)



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