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# Information-Seeking Preferences in Diverse Patients Receiving a Genetic Testing Result in the Clinical Sequencing Evidence-Generating Research (CSER) Study

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## Abstract

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Ethics Declaration

The study was approved by Institutional Review Boards (IRBs) at each CSER site as listed in Supplementary Table 1. Written informed consent was provided by adult participants ≥18 years of age, or by parents or legal guardians on behalf of their children <18 years of age or ≥18 years of age who were unable to consent independently. Assent was obtained from minors and intellectually disabled adults whenever possible.

Conflict of Interest

None of the authors have any conflicts to disclose.

**Purpose:** Accurate and understandable information following genetic testing is critical for patients, family members, and professionals alike.

**Methods:** As part of a cross-site study from the Clinical Sequencing Evidence-Generating Research (CSER) consortium, we investigated the information-seeking practices among patients and family members at five to seven months after genetic testing results disclosure, assessing the perceived utility of a variety of information sources, such as family and friends, healthcare providers, support groups, and the internet.

**Results:** We found that individuals placed a high value on information obtained from genetics professionals and healthcare workers, independent of genetic testing result case classifications as positive, inconclusive, or negative. The internet was also highly utilized and ranked. Study participants rated some information sources as more useful for positive results compared to inconclusive or negative outcomes, emphasizing that it may be difficult to identify helpful information for individuals receiving an uncertain or negative result. There were few data from non-English speakers, highlighting the need to develop strategies to reach this population.

**Conclusion:** Our study emphasizes the need for clinicians to provide accurate and comprehensible information to individuals from diverse populations following genetic testing.

### Keywords

Information-seeking preferences; genetic testing; diverse populations; exome sequencing; genome sequencing

## Introduction

Advances in genomic technology and improved access to genetic testing have increased the number of patients and families that obtain positive or probable positive results following genomic sequencing.[1] Ending the diagnostic odyssey, however, can generate a need for information on newly identified conditions. Following the return of a positive or probable positive genetic testing result, contact with genetics professionals may be limited and therefore related follow-up care may require the involvement of non-genetics healthcare professionals.[2] Inconclusive or negative genetic testing results may also elicit information seeking, Accurate and comprehensible information sources are therefore critical to improve understanding for patients and families when access to geneticists and genetic counselors is minimal.

Previous research has demonstrated that patients use a variety of sources to increase understanding and knowledge and to gather information before and after counseling in addition to receiving guidance from a genetics consultation.[3] Many patients seek information online and from other family members, but some individuals who have. or are at risk for, a genetic condition often find it difficult to ascertain trustworthy and understandable facts.[3] A previous study showed that health information seekers who first consult the internet often have more educational experience, and have a higher income than individuals who initially turn to other sources, including family members, handouts from hospitals, and media including newspapers/radio/television and magazines.[3] In the Health Information National Trends Survey (HINTS) study of primary information sources amongst a diverse

population of adults from the United States, a greater percentage of participants reported using the internet as their first source compared to family/friends/co-workers, health care professionals, and traditional media.[4] There was no association between race/ethnicity and health information seeking on the internet, although greater educational experience, higher socioeconomic status, younger age, and having appropriate skills were positively associated with internet use.[4,5] In contrast, older age, Hispanic ethnicity, low socioeconomic status, and low internet skills were determinants for using a health care provider or traditional media sources that may become dated more quickly, such as print and magazines. Patients who used the internet also preferred numbers rather than words to describe risk and considered it very important to have electronic access to personal medical information.[6] In a prior study of information-seeking behaviors amongst 185 underserved individuals, seeking health information online was infrequent.[7] Another study found that 71% (54/76) Latino participants who predominantly spoke Spanish did not seek online health information for themselves or others.[5]

The lack of knowledge regarding information-seeking preferences after genetic testing is exacerbated by a relatively limited amount of data on diverse populations. There is an urgent need to better understand the preferences of these groups, given the lack of research efforts in this area and the existing disparities in clinical care. An improved understanding of health decision-making in medically underserved groups that are underrepresented in genomics research is critical for promoting health equity.[8] The Clinical Sequencing Evidence-Generating Research (CSER) consortium has investigated the clinical utility of exome and genome sequencing in the care of individuals from diverse populations, which have been historically underserved and underrepresented in medical research, at six sites across the United States.[9] As part of a cross-site, collaborative research study for this consortium, we describe the information-seeking practices among patients and family members at five to seven months after genetic testing results disclosure.

## Materials and Methods

### Study Sites

Survey data related to information-seeking preferences was collected by five of the sites participating in the CSER consortium – the Program in Prenatal and Pediatric Genomic Sequencing ($P^3$EGS) project, the NYCKidSeq project, the SouthSeq project, the NCGENES 2 project, and the KidsCanSeq project. A brief synopsis for each study, including the patient populations studied and languages used, care site(s), genomic technologies, variant interpretation pipelines, and practices for results disclosure, is provided in Supplementary Table 1. Each project obtained informed consent according to an institutional review board (IRB)-approved protocol (Supplementary Table 1). In keeping with the recruitment goals of the CSER study, all sites aimed to enroll a minimum of 60% of patients from underserved and under-represented populations. These underserved and under-represented populations were broadly categorized according to the definition of “medically underserved” provided by the Health Resources and Services Administration (HRSA) that identifies geographic areas and populations with a lack of access to primary care services in the United States.[9]

## Demographic Data and Harmonized Measure Data for Information Sources

The CSER sites utilized harmonized survey measures that were created by study investigators.[10] We designed a survey to study the information-seeking preferences of patients and families who received genetic testing results with a case classification of definite positive, probable positive, inconclusive, or negative, The CSER-wide definitions for these case classifications are given in Supplementary Table 2. Definitive positive and probable positive case classifications were analyzed as positive results, whereas inconclusive and negative case classifications were considered as inconclusive and negative results respectively. The survey (Supplementary Table 3) was administered in person, by telemedicine, or by email at five to seven months after the return of a genetic testing result. Study participants were asked whether the following information sources were consulted after genetic testing results were disclosed to the patient or family member(s): 'Family and Friends', 'Facebook', 'Support groups', 'My/my child's other doctors' (hereafter referred to as 'My/my child's other healthcare providers'), 'Internet search' (abbreviated to 'Internet'), 'Books and other printed media' (abbreviated to 'Books and printed media'), and 'Information provided by the doctor (hereafter referred to 'My/my child's genetics provider') who ordered my/my child's genetic testing', and 'Other'. The perceived usefulness of each information source was rated on a Likert scale from 1 ('not useful at all') to 5 ('highly useful'). Patients and families who did not respond were provided with up to 3-6 reminders by email, telephone, or text; reasons for lack of response included participant withdrawal, survey refusal, or failure to return the survey. Further details are provided in Supplementary data.

In addition to data from the survey, harmonized demographic data was available for CSER study patients, enabling us to collate information regarding the estimated yearly income per patient or family household, the educational experience, and the preferred language of the patient or parent/guardian completing the questionnaire. One family member of the patient completed the survey for all patients under 18 years of age; unfortunately, demographic data from participants completing the surveys were not available.

## Data Analysis

Data download, conversion of categories to numerical data and methods for data analysis are described in Supplementary data. We collected the total number of participant responses for each information source, and as participants answered independently for each source, they had the option to check that they had used just one information source, or to check that they had used multiple sources. We analyzed the data in two ways: as data from all five sites combined, and as data separated according to individual study sites. Similarly, we examined the data for income level and educational experience as combined data from all five sites and as data from each individual site. As the results classifications (positive, inconclusive, or negative) could affect the perceived usefulness of the information sources, we examined the data for perceived utility, income level, and educational experience for each different result classification. For this analysis, we included positive results from three sites ($P^3$EGS, NYCKidSeq, and SouthSeq) and inconclusive and negative results from two sites (NYCKidSeq and SouthSeq), omitting the two sites (NCGENES 2 and KidsCanSeq) with smaller numbers that contributed less than 5% of the data to this analysis.

### Interviews

All families enrolled in the P[3]EGS study at UCSF were eligible to participate in semi-structured interviews exploring their experiences of exome sequencing.[11] This methodology is described in Supplementary data.

### Statistical analysis

Descriptive statistics for each set of data were run in Excel. An analysis of variation (ANOVA) test was performed to assess statistical significance between the means for participant income level and educational experience for each information source, across different sites together and by case status. To test whether result type was associated with Likert responses, educational experience, and income, ANOVA was used. For pairwise comparisons, t-tests assuming equal variance were performed to determine significance levels between individual datasets. Statistical significance was determined by a two-tailed, *P*-value <0.05; t-tests were also used to compare data for perceived utility, income level, and educational experience.

To evaluate whether the Likert scores for the information sources differed, we used linear regression with individual as a random effect with the covariance structure set to unstructured to account for potential within subject correlation. To ensure that differences in Likert scores of the information sources were not driven by result type or site, we used linear regression. In our regression model, the dependent variable was the Likert score with the independent variable being the type of information and result type included as a covariate. Site and individual were included as random effects, with the covariance structure set to unstructured. To verify that associations between result type and Likert responses, income, and educational experience were independent, we performed a multivariable nominal logistic regression, including all factors as predictors jointly. Regression models were run in JMP v16.2.

## Results

### Use of information sources

The numbers of survey participants who used one or more of the information sources after receiving a genetic testing result is shown for each site in Figure 1 and a summary of data for the survey respondents is provided in Supplementary Table 4. Amongst the participants who did use one or more information sources, there was a mean of 2.82 sources used, with a standard deviation of 2.01, a median of 2 and interquartile range 1-4. The total number of entries for all information sources ranged from 117 (KidsCanSeq) to 721 (NYCKidSeq) for the different CSER sites. The data from SouthSeq, NCGENES 2, and KidsCanSeq showed a relatively even distribution of the numbers of participants who consulted each information source, whereas the charts from NYCKidSeq and P[3]EGS showed an uneven distribution, including larger percentages of participants utilizing the ‘Internet’ (for example, 31.7% from the P[3]EGS study) and ‘My/My child’s other healthcare providers’ (23.4% from NYCKidSeq). Smaller percentages of participants from these sites consulted ‘Books and printed media’ (1.5% of entries from NYCKidSeq and 3.8% from P[3]EGS).

### Perceived utility of information sources – combined and site-specific data

Perceived usefulness for each information source was assessed with a Likert scale ranging from 1 (‘not useful at all’) to 5 (‘highly useful’). This analysis included data from different genetic tests and all types of results. The data was analyzed as the mean for each information source for all sites (Figure 2, Supplementary Table 5) and for each of the separate sites (Supplementary Tables 6A-E). For the data from all sites combined, information source differed in their perceived usefulness ($P < 0.0001$) after accounting for individual as a random effect, indicating substantial variation in levels of satisfaction with different information sources. The perceived utility of the information sources was still significantly different after including the type of result as a covariate and site and individual as random effects ($P < 0.0001$). The two information sources that were most highly ranked for the combined data were ‘My/my child’s genetics provider’ (mean 4.16; Supplementary Table 5) and ‘My/my child’s other healthcare providers’ (mean 4.10) with no significant difference in perceived usefulness between these two information sources ($P = 0.16$). The perceived utility for ‘My/my child’s genetics provider’ was significantly higher than for most other information sources (Figure 2). These results emphasize that patients perceive significant benefit from information obtained from both their genetics provider and other members of their healthcare team. The ‘Internet’ (mean 3.79) was rated below ‘My/my child’s genetics provider’ and ‘My/my child’s other healthcare providers’, but above the other information sources. The data for perceived usefulness at each separate CSER site also showed that NYCKidSeq, SouthSeq, and KidsCanSeq participants rated ‘My/my child’s genetics provider’ and ‘My/my child’s other healthcare providers’ as having the greatest utility (Figure 2; Supplementary Tables 6B, 6C and 6E).

### Income level and educational experience – combined and site-specific data

There was a strong positive correlation between educational experience and income ($P < 0.0001$), but not between case status and income or case status and educational experience (Supplementary Table 7). We examined the mean income level in study participants who used information sources as combined data from all sites (Supplementary Table 5) and as data from each of the individual sites (Supplementary Tables 6A-E). The combined data showed that the information source associated with the highest income level was ‘Internet’, with a mean income level of 4.02 that corresponds to approximately $80,000 (Supplementary Table 5). The mean income level for participants who used the ‘Internet’ was significantly higher than for ‘Books and printed media’ (mean 3.38; $P < 0.05$) and significantly higher than for ‘Support groups’ (mean 3.51; $P = < 0.05$) but was not significantly increased compared to the mean income for the remaining information sources. There were no significant differences between the mean educational experience and perceived usefulness of information sources, either within individual projects or among all sites combined.

### Perceived utility of information sources according to result type – combined and site-specific data

We next examined the perceived utility of the information sources according to the result classification as positive, inconclusive, or negative (Supplementary Table 8). Combining the

data for positive results from three sites (Table 1), we found that there was a significant difference in perceived usefulness for the information sources surveyed ($P = 0.008$), with the highest satisfaction given to 'My/my child's genetics provider' (mean 4.29) and 'My/my child's other healthcare providers' (mean 4.15; Figure 3). This result is consistent with the data for all types of results combined. Perceived utility was significantly different for the different information sources for inconclusive and negative results ($P = 0.0005$ and $< 0.0001$, respectively; Table 1). In particular, 'My/my child's genetics provider', 'My/my child's other healthcare providers', 'Internet', and 'Family and friends' were considered to have higher utility for inconclusive results than 'Support groups', 'Books and printed media', and 'Facebook' (Table 1; Figure 3). Similarly, for negative results, 'My/my child's genetics provider', 'Internet' and 'My/my child's other healthcare providers' were also perceived to be more useful compared to 'Books and printed media', 'Support groups', and 'Facebook' (Table 1; Figure 3).

Comparing perceived utility across the different result types for single information sources showed that utility declined for most information-sources when an inconclusive result was received compared to obtaining a positive result (Table 1; Figure 3). Study participants rated 'Facebook' as most helpful after receiving a positive result ($P = 0.002$) compared to other result types using a multivariable analysis adjusting for income and educational experience (Supplementary Table 9). Participants also found 'Support groups' were more helpful after a positive compared to a negative result ($P = 0.04$), but the result type was no longer significant after including income and educational experience in the model ($P = 0.14$). However, it is noteworthy that 'Support groups' was highly ranked for positive results (mean 4.02), supporting the establishment of patient, family and provider groups that can provide accurate information for rare genetic conditions. Perceived utility for single information sources did not demonstrate significant differences at the level of individual sites (Supplementary Tables 10A, 11A, and 12A). 'My/my child's genetics provider' (means 4.29, 4.24, and 3.97) and 'My/my child's other healthcare providers' (means 4.15. 4.13 and 4.18) retained the two highest rankings for all result types (Table 1), consistent with the results from our first analysis that were not subdivided according to result type.

## Income level and educational experience according to result type – combined and site-specific data

We examined income level and educational experience according to result type for both combined and site-specific data. The combined data showed that income level was lower for participants who used 'Facebook', 'Books and printed media', and 'My/my child's genetics provider who ordered the genetic testing' after a negative result (Table 1B; Supplementary Tables 10B, 11B and 12B; Figure 4), and these significant differences persisted after adjusting for educational experience and the Likert score of the information source (Supplementary Table 9). Similar findings were not observed for site-specific analyses and the significance of this result is uncertain. Despite significant variation for some information sources, we also could not make definite conclusions regarding variation in educational experience with different case classifications (Table 1C; Supplementary Tables 10C, 11C and 12C; Figure 5).

### Perceived utility for information sources In respondents who used more than one information source

A linear regression for a pairwise comparison of information sources with individual as a random effect showed significant differences in perceived utility between different information sources when used by the same individual (Supplementary Table 13).

### Information-seeking by preferred language

Participants completing the surveys were predominantly English speakers with very few participants preferring ‘Another language’ or ‘Equally comfortable in English and another language’. The small numbers precluded analysis for all sites except NYCKidSeq and this data is shown in Supplementary Table 14. Almost all participants who selected ‘Another language’ were Spanish speaking (data not shown). Spanish-speaking participants preferred ‘My/my child’s other healthcare providers’ (mean = 4.63) compared to ‘My/my child’s genetics provider’ (mean = 4.29), ‘Internet’ (mean = 3.82) and ‘Family and friends’ (mean = 3.79). For this group, ‘Facebook’ and ‘Books and printed media’ were not consulted and only one participant used ‘Support groups’.

### Interview findings

The interview data are summarized in Supplementary data. Participants revealed that information-seeking was helpful in determining questions for healthcare providers and future directions for study participants (“We wait and as we study what we can online, we’re preparing a list of questions. And so at our next meeting, we’ll be able to ask intelligent questions that are going to help us out with [Proband].” The interview data indicated several reasons for not consulting information sources, including perception of a low risk for recurrence (“..like really, really low chances of the same thing happening again to us. So I didn’t do a lot more research since then”), a lack of actionability (“…If the experts have told me that there is nothing to help her now, what other information am I searching for?”) and a ‘wait and see’ approach (“They said that there might be something – we just haven’t studied it or we haven’t taken a look at it….”). Other barriers to information seeking included time (“No, because I work all day and I’m very busy so I haven’t had time for that”), language (“The thing is that as everything is in English it’s hard for me…”), expertise (“…And I have other friends in the medical field as well too. And so I ask them or I Google it.”), and social support (“Interviewer: Is there anyone who could help you search more information?” Parent: “Honestly, I don’t know.”). Many providers advised caution for patients consulting the internet, which was heeded (“We try – at least I try not to indiscriminately Google…”), although curiosity often prevailed (“…after I was told about that I was told not to look in the pictures – it’s really hard not to.”). Others found that internet searches could result in stress (“…And I noticed that when I do look something up, it just gets me more stressed, so I try not to do that.”).

## Discussion

We surveyed the information-seeking practices of participants and family members after receiving genetic testing results from five projects in the CSER consortium. For all sites, participants and family members rated their healthcare professionals as highly valued

sources of information, supporting the perception of significant benefit from the information provided by clinicians who ordered the genetic testing and subsequently managed their care. This finding was apparent from data analyzed both as combined from all sites and separately and was independent of result type. This finding affirms the trusted role of healthcare professionals as information sources, but also emphasizes the need for healthcare professionals to provide appropriate and accurate information. The central role of the genetic counselor as an information source before and after genetic testing has previously been identified in quantitative studies, even though 25-35% of counselees sought additional information before or after result disclosure.[12,13] The results also suggest that healthcare providers should be taught best practices for obtaining and sharing reliable information about genetic conditions and rare diseases.

Our results showed varying preferences for different information sources across the CSER sites in terms of number of participants using each source. These differences may reflect the number of surveys completed and enrollment across sites, together with differences in study design that influenced diagnostic yield. For example, lower numbers of positive results were anticipated from germline genetic testing in pediatric patients with cancer compared to germline genetic testing in pediatric patients with neurodevelopmental disorders.[14] Although we had small numbers when analyzing the use of information sources according to result classification, perceived utility declined for most information sources after an inconclusive result compared to a positive result. Study participants rated ‘Facebook’ and ‘Support groups’ as most helpful after receiving a positive result in comparison to an inconclusive or negative result; in particular, ‘Support groups’ was highly ranked for positive results. These changes in rating are understandable, in that negative or inconclusive results offer less assistance for directing patients and families to specific resources. Although there is evidence that patients still value an uncertain or negative genetic testing result,[15,16] the lower rankings for information sources consulted after inconclusive or negative results may emphasize a lack of definitive information or explanation that is available for individuals who receive these types of result, or it may indicate that participants felt that less information was needed. Regardless, at the time of disclosure for these result categories, health professionals could consider proactively addressing this situation and providing guidance to patients and families who might consult information sources without a clear direction or encounter a paucity of information.

Consistent with prior work that showed that the internet was a popular source of health information,[17,18] our data also showed that ‘Internet’ was highly consulted and highly rated. Although ‘Internet’ was rated below ‘My/my child’s genetics provider’ and ‘My/my child’s other healthcare providers’, it was perceived as significantly more useful than other information sources. However, using the internet can result in accession of inaccurate information, in addition to the emotional stress of uncovering unexpected and unwanted knowledge by patients and families. The quotes from patient interviews provided indicate that both healthcare professionals and patients were aware of these possibilities.

The combined data from multiple sites showed that the information source associated with the highest income level was ‘Internet’, with a mean income level that was significantly higher than for ‘Books and printed media’ and ‘Support groups’ but that was not

significantly increased compared to the remaining information sources. This result was supported by the results from the $P^3$EGS and NYCKidSeq studies as single sites. There was no significant difference in educational experience for internet users in combined data or in data from the individual CSER sites.

Our survey respondents were predominantly English speakers and there was minimal data from participants who preferred a non-English language. Almost all participants who selected 'Another language' were Spanish-speaking. It is unclear if participants who preferred a non-English language did not consult more sources because of language barriers reducing the usefulness of these information sources or a preference for sources that were not surveyed. Our results suggest a need for more accessible information on genetic conditions to be available to non-English speakers. Strategies for families limited by language or expertise could include joint information-seeking together with an authoritative source to fill in knowledge gaps, confirm understanding, and to develop follow up questions. However, there are significant inequalities in the abilities of study participants to maximize their searching and gain sufficient knowledge to re-contact their clinical team. Our results also imply that it is likely that not all study participants who receive a genetic testing result will seek out the information sources that we surveyed.

There are several limitations to this study. Some sites surveyed participants receiving an inconclusive or a negative result, whereas other sites did not; we attempted to control for this difference by analyzing results according to different results in addition to a combined analysis. The data collection period at five to seven months after results provision was relatively broad, thus enabling opinions regarding source usefulness to fluctuate and perhaps giving some participants more time than others to consult information sources. Additional limitations include a low response rate for some sites and statistical approaches that did not include all demographic characteristics. Lastly, the numbers of study participants using information sources was modest but may indicate a real preference for some sources over others, or lack of interest in using some of the information sources that we surveyed. The age of the patient or family member completing the survey was not available, thus we could not compare the use of information sources with the age of the survey respondents.

In summary, we surveyed the information-seeking preferences of diverse participants who received a genetic testing result. The aggregated results showed that participants and family members considered their genetics professionals and other healthcare workers to be highly valued sources of information and this finding was valid independently of result classification as positive, inconclusive, or negative. The Internet was also highly utilized and highly ranked. Our results suggest several areas in which practice improvements could be considered. Study participants rated some information sources as more helpful for positive results compared to inconclusive or negative results and this finding implies that it can be challenging to provide relevant information for individuals receiving uncertain or negative genetic testing results. In addition, there were few data from non-English speakers, suggesting that the information sources that we surveyed may not have met their needs, highlighting the necessity of developing strategies to reach this population. Our results stress a critical need for healthcare providers to be trained in optimal practices for providing accurate and comprehensible information for genetic conditions and rare diseases.

## Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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## Data availability

Data is available from the authors on request.

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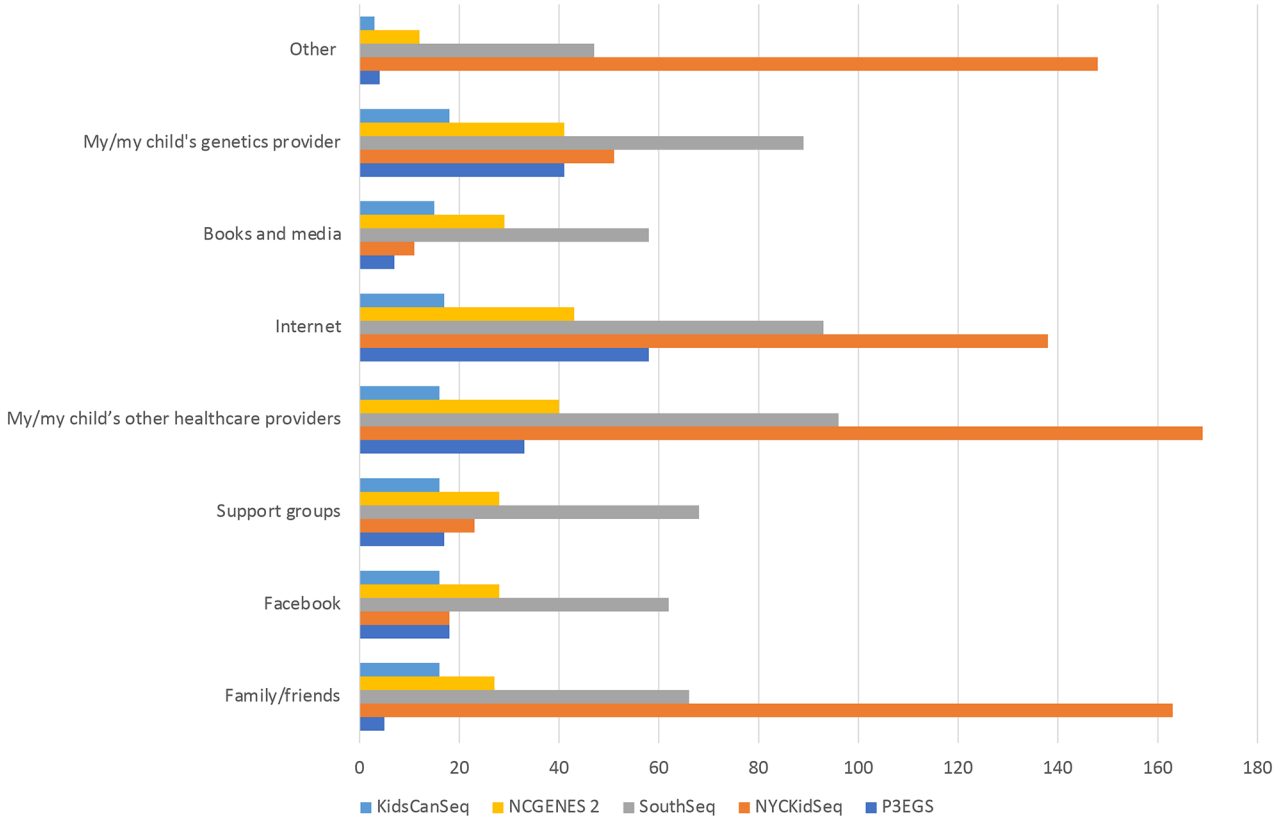


**Figure 1. Bar graphs showing information sources consulted after receiving a genetic testing result for participants enrolled in the Clinical Sequencing Evidence-Generating Research (CSER) consortium.**

Each bar graph shows the number of survey respondents who used the following information sources - 'Family and Friends', 'Facebook', 'Support groups', 'My/my child's other healthcare providers', 'Internet', 'Books and printed media', 'My/my child's genetics provider', and 'Other'. Data from Program in Prenatal and Pediatric Genomic Sequencing ($P^3EGS$) project, University of California San Francisco, is shown in dark blue, data from NYCKidSeq project, Icahn School of Medicine at Mount Sinai, is shown in orange, data from SouthSeq project, HudsonAlpha Institute for Biotechnology, is shown in grey, data from NCGENES 2, University of North Carolina, Chapel Hill, is shown in yellow, and data from KidsCanSeq project, Baylor College of Medicine, is shown in light blue.

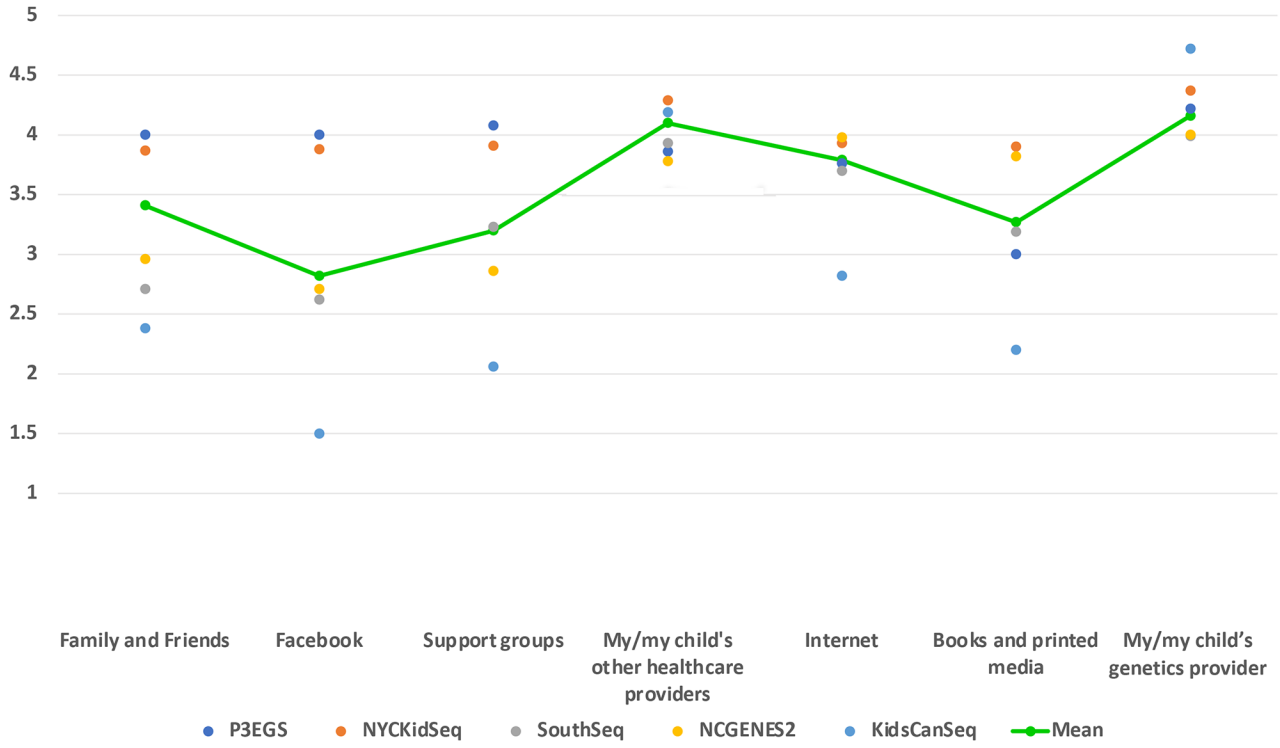


**Figure 2. Perceived usefulness of information sources consulted by participants enrolled in the Clinical Sequencing Evidence-Generating Research (CSER) study after receiving a genetic testing result.**

Each dot represents the mean of data derived from a Likert scale ranging from 1 to 5 on the Y-axis, with 5 representing 'very useful' to 1 representing 'not useful at all'. The data from all five CSER sites ($P^3$EGS, NYCKidSeq, SouthSeq, NCGENES 2 and KidsCanSeq) are shown for the information sources 'Family and Friends', 'Facebook', 'Support groups', 'My/my child's other healthcare providers', 'Internet', 'Books and printed media', and 'My/my child's genetics provider who ordered the test'. Data from Program in Prenatal and Pediatric Genomic Sequencing ($P^3$EGS) project, University of California San Francisco, is shown in dark blue, data from NYCKidSeq project, Icahn School of Medicine at Mount Sinai, is shown in orange, data from SouthSeq project, HudsonAlpha Institute for Biotechnology, is shown in grey, data from NCGENES 2, University of North Carolina, Chapel Hill, is shown in yellow, and data from KidsCanSeq project, Baylor College of Medicine, is shown in light blue. The mean for all sites combined is represented in green and these dots are connected for better visibility, although the data were not continuous. The dots demonstrate high perceived utility for 'My/my child's other healthcare providers' and 'My/my child's genetics provider who ordered the test'.

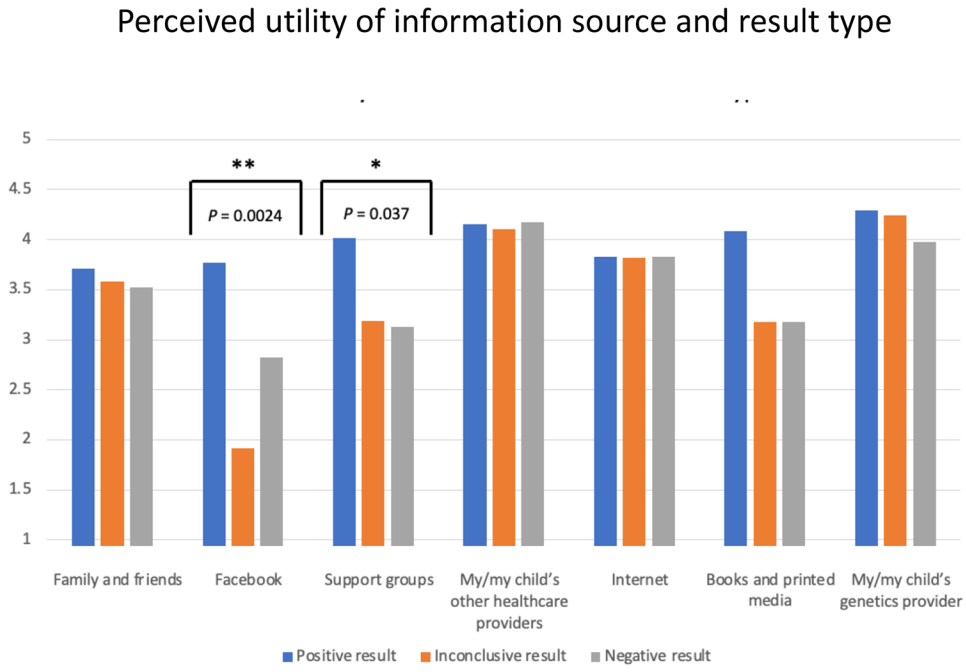


**Figure 3. Perceived utility of information sources and result type for participants enrolled in the Clinical Sequencing Evidence-Generating Research (CSER) study.**

The graph shows the mean perceived utility for data from three project sites - P$^3$EGS, NYCKidSeq, and SouthSeq. Perceived utility is shown with a Likert scale on the Y-axis ranging from 1 ('not useful at all') to 5 ('highly useful') for each information source. Three different result types (positive, inconclusive, and negative) are shown, with the means for positive results shown in blue, the means for inconclusive results shown in orange, and the means for negative results shown in grey. Significance is marked with * ($P < 0.05$), ** ($P < 0.01$), and *** ($P < 0.001$).

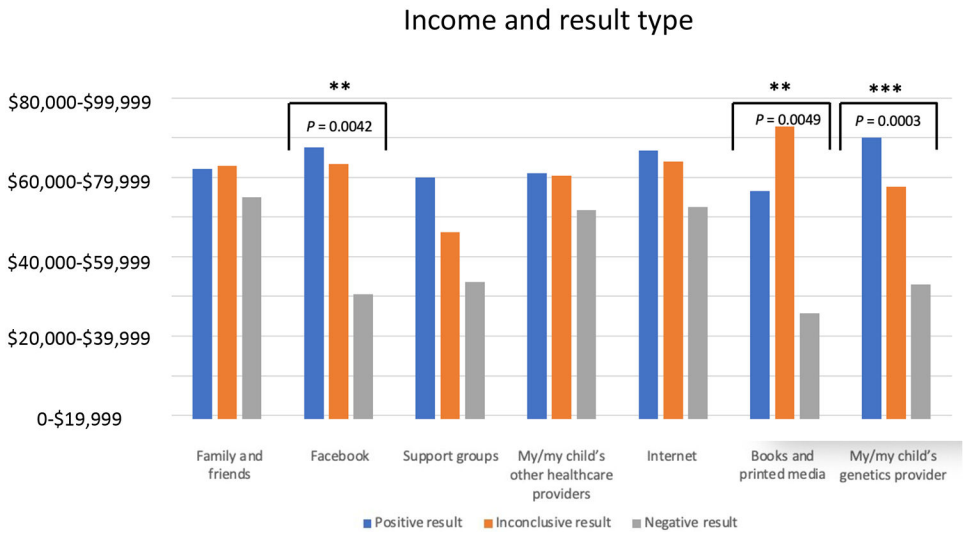


**Figure 4. Income and result type for participants enrolled in the Clinical Sequencing Evidence-Generating Research (CSER) study.**

The graphs show the mean income converted to scale (see Supplementary data for methodology) for result type (positive, inconclusive, and negative) for three project sites - P$^3$EGS, NYCKidSeq, and SouthSeq. The numbers on the Y axis corresponds to household income as follows: 0-$19,999, $20,000-$39,999, $40,000-$59,999, $60,000-$79,999, and $80,000-$99,999 (see Supplementary data). The means for household income for positive results are shown in blue, the means for inconclusive results are shown in orange, and the means for negative results are shown in grey. Significance is marked with * ($P < 0.05$), ** ($P < 0.01$), and *** ($P < 0.001$).

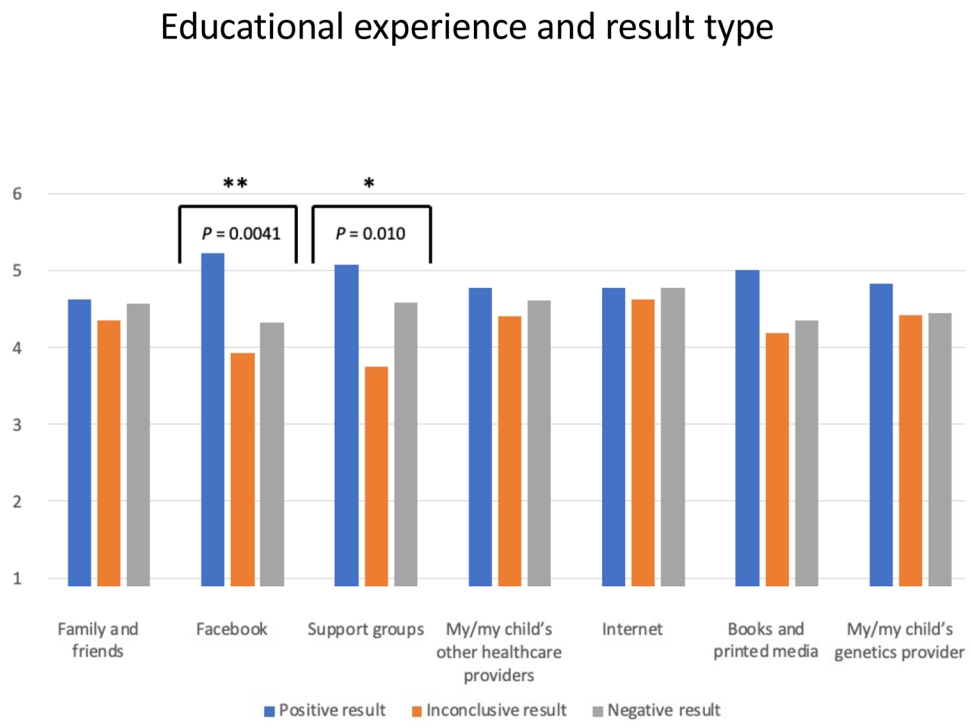


**Figure 5. Educational experience and result type for participants enrolled in the Clinical Sequencing Evidence-Generating Research (CSER) study.**

The graphs show the mean educational experience converted to scale for result type (positive, inconclusive, and negative) for three project sites - $P^3$EGS, NYCKidSeq, and SouthSeq. The numbers on the Y axis corresponds to educational experience as follows: 1 = Less than high school; 2 = Attended high school but did not receive a diploma; 3 = High school diploma/GED; 4 = Some post high school education; 5 = Associate college degree, occupational, technical, or vocational program, degree, or certificate; 6 = Bachelor's degree; 7 = Graduate or professional degree, for example, Master's degree, doctoral degree, MD and other (see Supplementary data). The means for educational experience for positive results are shown in blue, the means for inconclusive results are shown in orange, and the means for negative results are shown in grey. Significance is marked with * ($P < 0.05$), ** ($P < 0.01$), and *** ($P < 0.001$).

## Table 1

Combined data for information source use from P[3]EGS, NYCKidSeq, and SouthSeq projects for positive, inconclusive, and negative genomic testing results

| Information Source | *n* | Positive Result | *n* | Inconclusive Result | *n* | Negative Result | *P* value |
|---|---|---|---|---|---|---|---|
| Perceived utility of information source | | | | | | | |
| Family and friends | 43 | 3.63 +/− 1.50 (0.23)[a] | 92 | 3.6 +/− 1.53 (0.16) | 92 | 3.55 +/− 1.47 (0.15) | 0.96 |
| Facebook | 43 | 3.77 +/− 1.38 (0.21) | 13 | 2.15 +/− 1.82 (0.5) | 34 | 2.82 +/− 1.75 (0.3) | 0.0024** |
| Support groups | 41 | 4.02 +/− 1.15 (0.18) | 17 | 3.29 +/− 1.99 (0.48) | 41 | 3.17 +/− 1.67 (0.26) | 0.037* |
| My/my child's other healthcare providers | 88 | 4.15 +/− 1.00 (0.11) | 89 | 4.13 +/− 1.24 (0.13) | 108 | 4.18 +/− 1.17 (0.11) | 0.97 |
| Internet | 122 | 3.85 +/− 1.23 (0.11) | 71 | 3.83 +/− 1.40 (0.17) | 80 | 3.84 +/− 1.25 (0.14) | 0.99 |
| Books and printed media | 25 | 3.52 +/− 1.26 (0.25) | 12 | 3.33 +/− 1.87 (0.54) | 34 | 3.18 +/− 1.53 (0.26) | 0.69 |
| My/my child's genetics provider[b] | 73 | 4.29 +/− 1.03 (0.12) | 37 | 4.24 +/− 1.28 (0.21) | 60 | 3.97 +/− 1.3 (0.17) | 0.27 |
| *P* value | — | 0.0079 | — | 0.0005 | — | < 0.0001 | — |
| | | | | | | | |
| Income Level | | | | | | | |
| Family and friends | 38 | 4.05 +/− 2.34 (0.38) | 85 | 4.15 +/− 2.74 (0.30) | 87 | 3.75 +/− 2.61 (0.28) | 0.58 |
| Facebook | 40 | 4.38 +/− 2.55 (0.40) | 12 | 4.17 +/− 2.79 (0.81) | 34 | 2.53 +/− 2.02 (0.35) | 0.0042** |
| Support groups | 38 | 4.00 +/− 2.56 (0.41) | 16 | 3.31 +/− 2.50 (0.62) | 41 | 2.83 +/− 2.39 (0.37) | 0.12 |
| My/my child's other health care providers | 78 | 4.05 +/− 2.42 (0.27) | 84 | 4.07 +/− 2.71 (0.30) | 102 | 3.59 +/− 2.55 (0.25) | 0.35 |
| Internet | 111 | 4.34 +/− 2.54 (0.24) | 65 | 4.20 +/− 2.67 (0.33) | 78 | 3.63 +/− 2.63 (0.30) | 0.17 |
| Books and printed media | 23 | 3.83 +/− 2.62 (0.55) | 11 | 4.64 +/− 3.04 (0.92) | 34 | 2.29 +/− 1.66 (0.28) | 0.0049** |
| My/my child's genetics provider | 66 | 4.50 +/− 2.72 (0.34) | 33 | 3.88 +/− 2.64 (0.46) | 57 | 2.65 +/− 2.07 (0.27) | 0.0003*** |
| *P* value | — | 0.86 | — | 0.91 | — | **0.0023** | — |
| | | | | | | | |
| Educational Experience | | | | | | | |
| Family and friends | 43 | 4.72 +/− 1.79 (0.27) | 91 | 4.33 +/− 1.85 (0.19) | 92 | 4.59 +/− 1.92 (0.20) | 0.46 |
| Facebook | 43 | 5.23 +/− 1.56 (0.24) | 13 | 3.85 +/− 1.52 (0.42) | 34 | 4.32 +/− 1.43 (0.25) | 0.0041** |
| Support groups | 41 | 5.07 +/− 1.59 (0.25) | 17 | 3.71 +/− 1.65 (0.4) | 41 | 4.61 +/− 1.43 (0.22) | 0.010* |
| My/my child's other health care providers | 88 | 4.78 +/− 1.77 (0.19) | 88 | 4.42 +/− 1.80 (0.19) | 108 | 4.61 +/− 1.87 (0.18) | 0.42 |
| Internet | 122 | 4.78 +/− 1.79 (0.16) | 71 | 4.58 +/− 1.86 (0.22) | 80 | 4.80 +/− 1.77 (0.20) | 0.70 |
| Books and printed media | 25 | 5.00 +/− 1.66 (0.33) | 12 | 4.08 +/− 1.51 (0.43) | 34 | 4.35 +/− 1.41 (0.24) | 0.15 |
| My/my child's genetics provider | 73 | 4.86 +/− 1.83 (0.21) | 36 | 4.33 +/− 1.77 (0.30) | 60 | 4.40 +/− 1.66 (0.21) | 0.20 |
| *P* value | — | 0.76 | — | 0.58 | — | 0.77 | — |

[a] All data are expressed as mean +/− standard deviation with the standard error in parentheses.

[b] My/my child's genetics provider = My/my child's genetics provider who ordered my/my child's genetic testing. Numbers for Income are lower than for perceived utility and educational experience due to some participants choosing not to share this data. The *P* values in the right-hand column were derived from analysis of variance, comparing the values between result types. The *P* values on the bottom row of perceived utility were derived from a linear regression with individual as a random effect and information source as the independent variable. The *P* values for income and education bottom rows were derived from analysis of variance. Significance is marked with *($P<0.5$), **($P<0.01$), ***($P<0.001$).