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ORIGINAL ARTICLE

Familial communication and cascade testing following elective genomic testing

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Abstract

Familial communication of results and cascade genetic testing (CGT) can extend the benefits of genetic screening beyond the patient to their at-risk relatives. While an increasing number of health systems are offering genetic screening as an elective clinical service, data are limited about how often results are shared and how often results lead to CGT. From 2018 to 2022, the Sanford Health system offered the Sanford Chip, an elective genomic test that included screening for medically actionable predispositions for disease recommended by the American College of Medical Genetics and Genomics for secondary findings disclosure, to its adult primary care patients. We analyzed patient-reported data about familial sharing of results and CGT among patients who received Sanford Chip results at least 1 year previously. Among the patients identified with medically actionable predispositions, 94.6% (53/56) reported disclosing their result to at least one family member, compared with 46.7% (423/906) of patients with uninformative findings ($p < 0.001$). Of the patients with actionable predispositions, 52.2% (12/23) with a monogenic disease risk and 12.1% (4/33) with a carrier status reported that their relatives underwent CGT. Results suggest that while the identification of monogenic risk during elective genomic testing motivates CGT in many at-risk relatives, there remain untested at-risk relatives who may benefit from future CGT. Findings identify an area that may benefit from increased genetic counseling and the development of tools and resources to encourage CGT for family members.

KEYWORDS
cascade testing, elective genomic testing, familial communication, familial impact, genetic counseling, primary healthcare

1 | INTRODUCTION

The utility of sharing genetic results for highly actionable conditions with at-risk relatives is well established (Committee on Gynecologic Practice, 2018; Roberts et al., 2018), and allows family members to benefit from targeted prevention, early detection, and personalized treatment of disease. This communication can drive cascade genetic testing (CGT), a targeted and effective strategy for identifying family members who may have the same genetic variants (Cornel & van El, 2017). Experts have argued that the wider implementation of CGT would lead to the identification of individuals with genetic conditions such as hereditary cancer syndromes more quickly and less expensively than population screening initiatives (Offit et al., 2020). Unlike other countries such as the Netherlands (Umans-Eckenhausen et al., 2001), the United Kingdom (Tosi et al., 2007), and Spain (Rubio-Marín et al., 2018), who have established systematic CGT programs to directly engage with patients and their family members, the United States relies heavily on a family-mediated model where the responsibility of informing relatives rests with the patient alone (McGowan et al., 2021; Srinivasan et al., 2020; Stefka et al., 2023). This increases the importance of understanding familial disclosure as a facilitator for CGT.

Previous studies have demonstrated that upon learning of their own medically actionable genetic findings, patients often share information about their result with at least one family member (Cheung et al., 2010; Finlay et al., 2008; Taber et al., 2015; Wynn et al., 2022). A recent publication reported that an average of 1.5 relatives per patient underwent CGT following familial disclosure (Stefka et al., 2023). Depending on the health system, clinical specialty, costs, and other factors that have previously been reported, uptake of CGT following familial disclosure varies widely from 8% to 94% (Cernat et al., 2021; Menko et al., 2019; Stefka et al., 2023). Most studies generating these data focus on patients whose testing was prompted by a personal or family history of a disease (Caswell-Jin et al., 2019; Conley et al., 2020; Elrick et al., 2017; Koehly et al., 2009). Less is known about disclosure and CGT when disease predispositions are identified through elective genomic testing programs (population genetic screening programs in which the test was not ordered for a clinical indication). Geisinger is one health system that has begun sharing data regarding family sharing and CGT following elective genomic testing in a primary care setting (Campbell-Salome et al., 2021; Schmidlen et al., 2022). Beyond what has been shared by Geisinger and a handful of other health systems implementing elective genomic testing, there is limited knowledge about family information sharing practices and uptake of CGT when these services are offered through a clinical, health system-wide program rather than a controlled research study (Wynn et al., 2022).

We addressed this evidence gap by summarizing communication of results and CGT following elective genomic testing provided to primary care patients in another major health system, Sanford Health. Beginning in 2018, the Sanford Health system offered the Sanford Chip, a service that provided pharmacogenomic and optional genetic risk information to adult patients across the health system (Christensen et al., 2021; Hajek et al., 2022). This program presents a unique opportunity to assess the familial impact following elective genomic testing in a clinical setting. Drawing from patient-reported data, we discuss information sharing practices with family and the frequency of subsequent CGT that can identify at-risk relatives and facilitate earlier targeted disease management and/or prevention interventions. The purpose of this study is to provide real-world data about communication of results and CGT to inform the development of policies that maximize the benefits of identifying inherited predispositions during elective genomic testing.

What is known about this topic

The sharing of genetic results with family members facilitates cascade genetic testing to identify individuals at increased risk for genetic disease, allowing for early intervention and management.

What this paper adds to the topic

Of those who participated in a clinical elective genomic testing program, nearly all patients identified with medically actionable predispositions shared their results with at least one family member. However, many patients with monogenic risks reported that their relatives did not undergo cascade genetic testing, highlighting a need for increased genetic counseling and the development of tools to simplify and streamline cascade genetic testing processes.

2 | METHODS

2.1 | Sanford Chip program

Sanford Health, the largest rural non-profit health system in the United States, launched the Sanford Imagenetics Initiative in 2014 with the goal of accelerating the implementation of EGT into all aspects of patient care (Christensen et al., 2021). From 2018 to 2022, the initiative offered the Sanford Chip, an elective, clinical laboratory-developed genetic test available to adult primary care patients available for $49. The Sanford Chip provided panel pharmacogenomic testing and screened for actionable predispositions (pathogenic and likely pathogenic variants associated with conditions with established prevention options). Genes that were screened were those included on the American College of Medical Genetics and Genomics v2.0 secondary findings list, excluding *NF2* and *WT1* (Green et al., 2013; Kalia et al., 2017). Dominant, biallelic recessive, and carrier status for autosomal recessive conditions *MUTYH*-associated polyposis and Wilson disease were offered. Sanford Chip recipients could receive pharmacogenomic testing but decline to be screened for actionable

predispositions. Testing was performed primarily using an array-based platform, with positive findings confirmed via orthogonal approaches prior to disclosure (Christensen et al., 2021).

Adult patients were eligible to receive the Sanford Chip upon approval from a primary care provider at Sanford Health and if they had an active account on the Sanford MyChart patient portal linked to their electronic medical record. Patients who enrolled in the program responded to invitations electronically sent through their MyChart patient portal. After clinical consent and testing, pharmacogenomic and actionable predisposition findings were automatically integrated into the electronic medical records and released into the MyChart patient portal of Sanford Chip recipients. Irrespective of any previous genetic testing, all patients were screened for the same genes and conditions as outlined in Table S1. All patients were offered the opportunity to speak with a genetic counselor both before and after receiving their results. Patients who were positive for at least one variant associated with an actionable predisposition were contacted by a Sanford Health laboratory genetic counselor when their results were released and offered referrals to clinical genetics. Those who elected to see a genetic counselor were counseled on the benefits of CGT, when indicated. During these conversations, testing options and potential costs were provided for CGT.

2.2 | METRICS survey development and administration

Sanford Health partnered with the Genomes2People Research Program and the Harvard Pilgrim Health Care Institute to launch the Imagenetics METRICS Study in 2019 (Christensen et al., 2021). A multidisciplinary team developed a REDCap-based survey (Harris et al., 2009) to administer to patients who received Sanford Chip results at least 1 year prior that included items asking about communication of results to relatives and CGT.

2.3 | Eligible participants

Patients who were eligible for these surveys were first queried through MyChart messages for permission to be contacted via their personal email address on file. Patients who consented to recontact were then sent the follow-up survey between January and December 2022. This study was approved by the Sanford Institutional Review Board and electronic consent was obtained from all participating patients for survey completion and review of medical records.

2.4 | Survey measures

At the beginning of the follow-up survey, each patient was asked if they had viewed their Sanford Chip results. Patients who reported either not having viewed their results or not remembering if they viewed their results did not receive questions pertaining to information sharing or CGT. This display logic was implemented to minimize confusion (e.g., implying patients should be sharing results they did not remember). All patients who reported having viewed their Sanford Chip results were asked which family members, if any, they shared their results with. Patients who received an actionable predisposition were also asked if any family members had CGT for the same variant, and if yes, which family members. The following categories of relatives were provided as a multiple select response for sharing and CGT: spouse/partner; children; siblings; parents; grandparent(s), grandchild(ren), aunt(s), uncle(s), niece(s), or nephew(s); other family members. Section instructions told patients that CGT items "are specifically about biological family members (related to you by blood)" except items about spouses or partners. Although not biologically related, spouses/partners were included as a category of relatives to elicit comprehensive data on family sharing practices, especially in the context of returning carrier results for recessive conditions. Patients were also asked questions on sociodemographic characteristics, as well as personal and family health history.

2.5 | Electronic medical record data

Demographic characteristics including patient age, gender, and Charlson Comorbidity Index score (Charlson et al., 1987) were collected from the patients' medical records. Details about the patients' enrollment in the Sanford Chip program, dates of disclosure, genetic findings, and provider visits were also captured from medical records. When patients did not self-report their race or ethnicity in the survey, these data were extracted from medical records.

2.6 | Statistical analysis

Patients who opted out of screening for actionable predispositions were omitted from analyses. The main analyses were restricted to patients who consented to and began the survey, agreed to screening for actionable predispositions, remembered viewing their results, and completed questions about sharing of results and CGT. We summarized patient characteristics with descriptive statistics, including counts with percentages, means with standard deviations, and medians with interquartile ranges where appropriate. Patient characteristics were compared between patients analyzed and those not analyzed (i.e., patients who did not remember viewing their results or did not complete questions about sharing of results and CGT) using Chi-squared tests or Fisher's exact test for categorical variables, Wilcoxon rank-sum or Kruskal–Wallis tests for ordinal variables, and *t*-tests or linear regression for continuous variables.

Analyses of family sharing and CGT were stratified by patients' risk status, including (a) "monogenic risk," when a pathogenic or

likely pathogenic variant was identified for an autosomal dominant condition; (b) carrier status, when a pathogenic or likely pathogenic variant was identified for an autosomal recessive condition; and (c) uninformative findings, when no pathogenic or likely pathogenic variants were identified. Given the small number of patients with informative findings, some analyses combined patients with monogenic risk findings and carrier status. Respondents who had both monogenic risk and carrier status findings were classified as having monogenic risk. Models that compared whether the likelihood of sharing results by type of relative varied by actionable predisposition finding type were conducted using generalized estimating equations (GEEs) to account for correlated responses within survey respondents. Independent variables included type of relative (spouse/partner, children, siblings, parents, second-degree relatives, and "other"), genetic status (monogenic risk, carrier status, and uninformative findings), and interaction terms between type of relative and genetic status. Models used logit linking functions and binomial distributions, and contrasts were used to compare whether the likelihood of sharing varied by familial relationship or by genetic status. Overall, model fit for GEEs was assessed as recommended using Corrected Quasi-likelihood under independence model criterion (QICC) values (Pan, 2001). Analyses of whether the likelihood of CGT differed between patients who had monogenic risk findings and carrier status findings used logistic regression. Three patients with monogenic risk findings and one patient with a carrier status finding were included in analyses, but reported separately and descriptively in analyses of information sharing and CGT, as they reported being aware of their monogenic risk or carrier status prior to receiving their Sanford Chip results.

Statistical significance was set at $p=0.05$. Exploratory analyses that examined whether the likelihood of sharing results and CGT varied by condition type of the variant (cancer, cardiovascular, or other) were omitted because of very small numbers. Analyses were conducted in R-4.3.0 (R Core Team, 2017).

3 | RESULTS

3.1 | Participant characteristics

Of the 11,028 eligible Sanford Chip patients who were asked for permission to be recontacted for the follow-up survey, 4324 (39.2%) agreed and were emailed a link to the follow-up survey (Figure 1). A total of 1887 patients (17.1% of eligible patients) consented and started the survey, and data from 962 patients (51.0% of survey initiators) were analyzed. Characteristics of Sanford Chip patients who provided data about the familial outcomes are summarized in Table 1. The majority of patients were female (66.3%) and non-Hispanic White (99.0%), and the median age was 56.1 years. Most patients (60.1%) had at least a college degree and 45% of patients reported a household income ≥$100,000. Nearly, two-thirds of patients were currently married (66.2%) and had an average of 1.8 children (SD = 1.3).

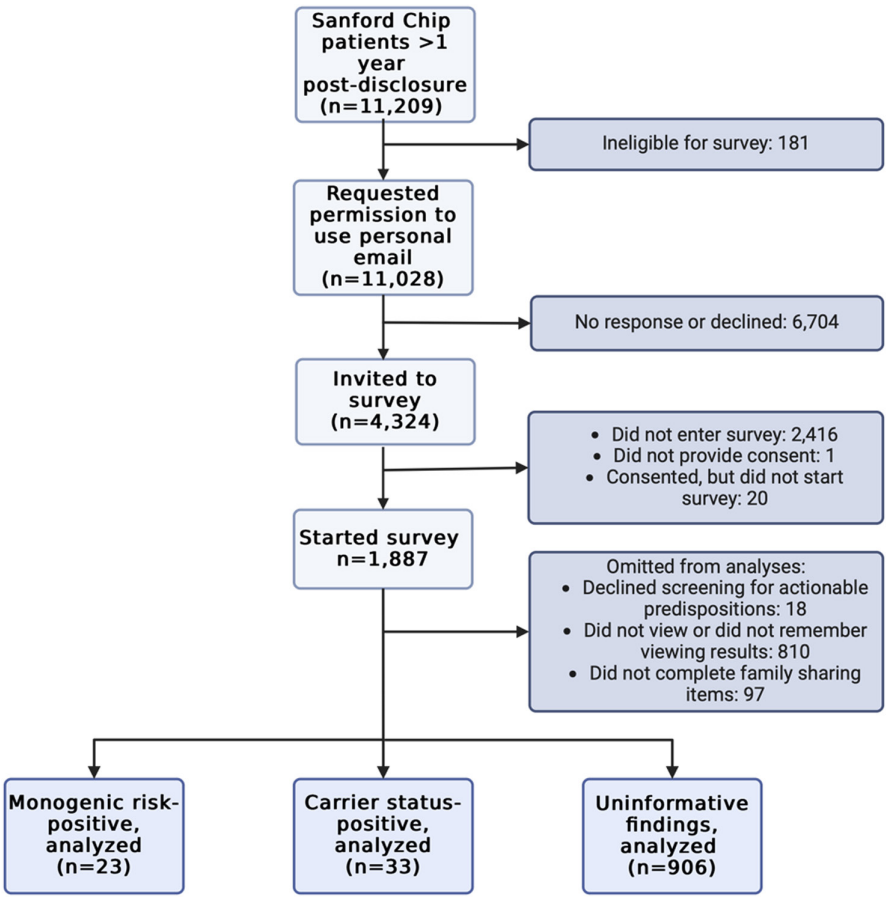

FIGURE 1 Flow process of survey administration and analysis.

TABLE 1 Characteristics of analyzed patients ($N = 962$).

Characteristic	N (%)
Genetic findings	56 (5.8%)
Positive for monogenic risk	23 (2.4%)
Positive for carrier status	35 (3.6%)
Sex[a]	
Female	632 (65.7%)
Male	330 (34.3%)
Median age (IQR)[a,b]	56.7 (23.6)
Non-Hispanic White	952 (99.0%)
Adopted	34 (3.5%)
Marital status	
Currently married	694 (72.1%)
Living with partner	48 (5.0%)
Mean number of children (SD)	1.8 (1.3)
Employment status[a]	
Full time employed	546 (56.8%)
Retired	302 (31.4%)
Household income	
<$50,000	135 (14.0%)
$50,000 to $99,999	283 (29.4%)
$100,000 to $149,999	219 (22.8%)
≥$150,000	215 (22.3%)
Missing income	110 (11.4%)
Educational attainment	
High school or less	59 (6.1%)
Post high school training or some college	285 (29.6%)
College graduate or more	616 (64.0%)
Missing education	2 (0.2%)
Mean Charlson comorbidity score (SD)[b]	1.6 (1.6)
Missing Charlson score	48 (5.0%)
Mean years since disclosure (SD)	2.8 (0.6)
Mean number of visits with a genetics specialist post-disclosure (SD)	0.2 (0.5)

Note: Survey data were analyzed for patients who had agreed to screening for medically actionable predispositions, remembered viewing their results, and completed items about sharing of results and cascade genetic testing.

[a]Among patients who initiated the survey and agreed to screening for medically actionable predispositions, a difference was observed between patients who were analyzed and patients who were not ($n = 907$) at $p < 0.01$.

[b]All characteristics were reported at time of survey completion except for age and Charlson comorbidity index score which were captured at time of Chip enrollment.

On average, patients completed the follow-up survey 2.8 years (range 1.1–4.0 years) after their Sanford Chip result disclosure date. Results disclosure dates ranged from May 20, 2018 to February 25, 2021 and survey completion dates ranged from January 17, 2022 to October 08, 2022. Fifty-six analyzed patients (5.8%) had actionable predisposition findings, including 23 (2.4%) who had monogenic risk findings and 35 (3.5%) with carrier status findings. Findings in specific genes are summarized in Table S2. One patient with an *LDLR* variant and one patient with a *TNNT2* variant were also each identified with carrier status for *MUTYH*. The four patients who knew about their findings previously were two patients with variants in *BRCA1*, one patient with a variant in *PMS2*, and one patient with carrier status in *MUTYH*.

Patients whose data were analyzed were more likely to be female, younger, and employed than patients who started the survey and agreed to actionable predisposition screening, but were not analyzed ($n = 907$) because they did not remember viewing their results or did not complete survey items about information sharing or CGT (all $p < 0.01$). Patients who did not remember viewing their results included five patients with monogenic risk, all of whom were either already being managed for the associated condition when results were disclosed or had follow-up with a genetics specialist following disclosure. Patients whose data were analyzed were also more likely to have actionable predisposition findings, had more post-disclosure visits with genetics specialists, were more educated, had lower Charlson Comorbidity Index scores, and had more time pass between results disclosure and survey completion (all $p < 0.01$). No differences were observed between patients with different categories of actionable predisposition findings (monogenic risk, carrier status, or uninformative findings; Table S3) except for visits with a genetics specialist and time between results disclosure and survey completion. Analyzed patients who had monogenic risk findings had an average of 1.3 follow-up visits with a genetics specialist following disclosure of results, compared with 1.1 visits for patients who had carrier status, and 0.1 visits for patients with uninformative findings ($p < 0.05$ in all pairwise comparisons). Moreover, among the analyzed patients, 91.3% (21/23) with monogenic risk findings, 93.9% (31/33) with carrier status, and 10.5% (105/906) with uninformative findings had at least one visit with a genetics provider. Analyzed patients with actionable predisposition findings also had less time pass between results disclosure and survey completion than analyzed patients with uninformative findings (2.5 vs. 2.8 years, on average; $p = 0.002$).

3.2 | Sharing practices

Patients with monogenic risk findings and patients with carrier status were more likely to report sharing results with at least one family member than patients with uninformative findings (100%, 90.6%, and 46.7%, respectively, both $p < 0.001$). Patients with monogenic risk findings were no more likely than patients with carrier status to report sharing results with at least one family member (100% vs. 90.6%, $p = 0.276$, Figure 2), but did report sharing results with more categories of relatives (2.9 vs. 2.0, $p = 0.002$). Analyses that examined information sharing in more detail showed that patients with monogenic risk findings or carrier status were more likely than patients with uninformative findings to disclose results with each type of relative except for other ($p < 0.001$ in all pairwise comparisons). Furthermore,

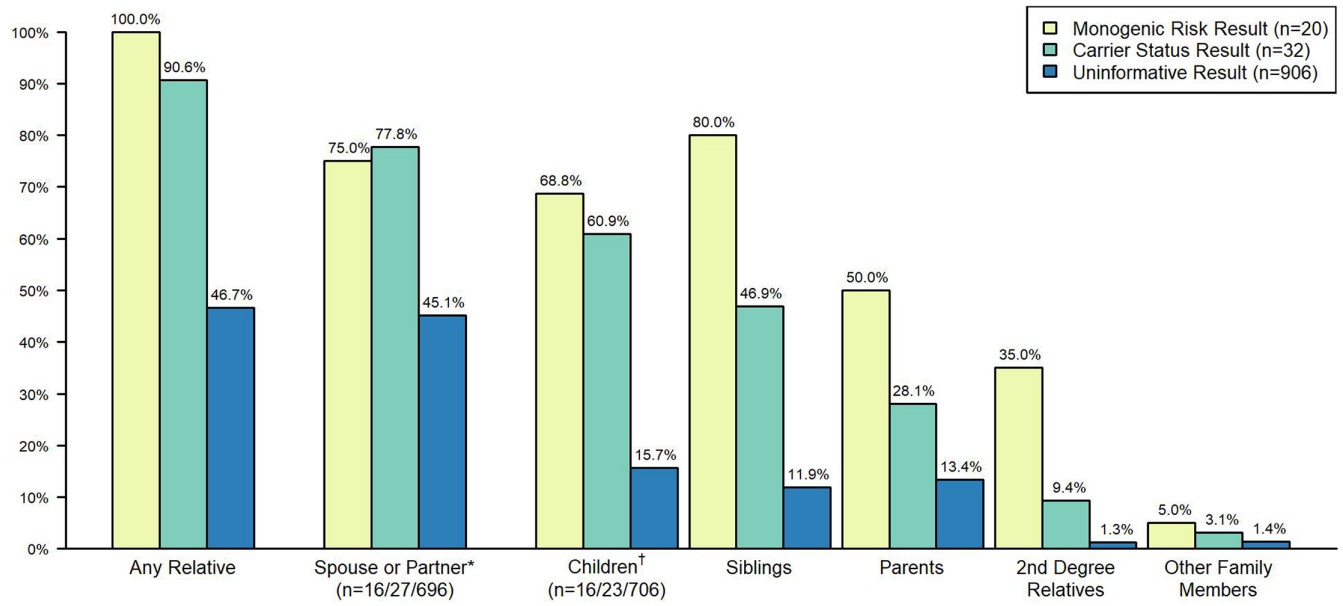

FIGURE 2 Sharing practices by result type. Bars represent the percentage of patients who shared their results for a disease predisposition with relatives. Percentages are calculated from the total number of patients who received the sharing item questions unless noted. Analyses omit patients ($n=4$) who were aware of their status for a medically actionable predisposition prior to disclosure of Sanford Chip results. *Analyses considered only patients who reported having spouses or partners. †Analyses considered only patients who reported having children.

TABLE 2 Comparison of cascade testing by result type (monogenic risk, carrier status) and at-risk relative type.

	Monogenic risk ($n=20$)	Carrier status ($n=32$)	p
Any family member	11 (55.0%)	3 (9.4%)	<0.001
Spouse or partner[a]	0 (0%)	0 (0%)	>0.999
Biological relatives	11 (55.0%)	3 (9.4%)	0.002
Children[b]	3 (18.8%)	2 (8.7%)	0.631
Siblings	7 (35.0%)	1 (3.1%)	0.003
Parents	3 (15.0%)	0 (0.0%)	0.052
Second-degree relatives	2 (10.0%)	0 (0.0%)	0.143
Other family members	0 (0%)	0 (0%)	>0.999

Note: Analyses omit four patients who were aware of their status for a medically actionable predisposition prior to disclosure of Sanford Chip results.

[a]Analyses considered only the 16 patients with monogenic risk results and 27 patients with carrier status results who reported having a spouse or partner.

[b]Analyses considered only the 16 patients with monogenic risk results and 23 patients with carrier status results who reported having at least one child.

patients with monogenic risk findings were more likely than patients with carrier status findings to share results with siblings (80.0% vs. 46.9%, $p=0.022$).

3.3 | Cascade genetic testing

Among patients who had not known they were positive for an actionable predisposition prior to disclosure of Sanford Chip results, patients were more likely to report that a biological relative received CGT for the same variant if the patient had monogenic risk findings rather than carrier status findings (55.0% vs. 9.4%, $p<0.001$, Table 2). Analyses of specific types of relatives suggest a greater likelihood of CGT among siblings when previously unknown positive findings were for a monogenic risk rather than carrier status (35.0% vs. 3.1%, $p=0.003$). One patient with a variant in *BRCA1* reported CGT in a sibling despite sharing results only with a spouse or partner. Among patients who had not known they were positive for an actionable predisposition prior to disclosure of Sanford Chip results, eight patients with monogenic risk findings (40.0%) and 25 patients with carrier status results (78.1%) specifically reported that none of their biological family members had

CGT because of their actionable predisposition finding ($p = 0.008$). Among the four patients who knew about their actionable predisposition previously, one reported CGT in a sibling motivated by Sanford Chip disclosure of a *BRCA1* variant, and another reported CGT in a sibling and second-degree relative motivated by Sanford Chip disclosure of *MUTYH* carrier status. No patients with actionable predispositions reported CGT for their spouses or partners. All participants who had carrier status findings and reported CGT ($n = 4$) were carriers of *MUTYH*-associated polyposis; none were carriers for Wilson disease.

4 | DISCUSSION

CGT offers a targeted approach for identifying individuals at increased risk of genetic disease, allowing for early intervention and management (Sermijn et al., 2016). This study is among the first to examine family information sharing and CGT from elective genomic testing offered in a clinical setting. Consistent with studies of CGT in high-risk populations (Caswell-Jin et al., 2019; Conley et al., 2020) and research populations (Wynn et al., 2022), the majority of patients with an actionable predisposition finding reported sharing their results with at least one family member, including all patients with monogenic risk findings. For both monogenic risk and carrier status patients, the most frequently informed at-risk relatives were first-degree relatives, followed by second-degree relatives. However, just over half of patients with monogenic risk findings and about one in eight patients with carrier status findings reported CGT in relatives. Results suggest that monogenic risk findings were shared more expansively within families than carrier status findings.

While it is encouraging to observe that all patients shared their monogenic risk results with at least one family member, several factors may have influenced the subsequent uptake of CGT observed through the Sanford Chip program, such as financial and logistical barriers for family members. While the Sanford Chip was a low-cost service and patients with an actionable predisposition were automatically referred to clinical genetic counseling, the cost of targeted CGT for their family members can be expensive. In addition, the coordination of such testing can often be complicated, especially if at-risk family members have not elected to see a genetic counselor (Choi et al., 2022; Srinivasan et al., 2020). Accessibility, affordability, and availability of CGT services are not unique problems to the Sanford Health system; finances (Campbell et al., 2017; van El et al., 2018), access to genetic testing services (Ulph et al., 2015), and the extra clinical referrals required to pursue CGT (van El et al., 2018) have all been previously cited as barriers to pursuing CGT services (Srinivasan et al., 2020). Insurance coverage is another commonly cited barrier to CGT (Campbell et al., 2017), both because insurers often do not cover this testing (Choi et al., 2022), and because of individuals' concerns over the potential for increased health, disability, and/or life insurance rates based on their genetic risk profile (Wurtmann et al., 2018).

Moreover, family dynamics (Lieberman et al., 2018; Stoffel et al., 2008) as well as perceptions of family members (Burns et al., 2016; Campbell et al., 2017; McClaren et al., 2013), attitudes toward genetic testing (Ormondroyd et al., 2014), privacy concerns (Stoffel et al., 2008), and levels of health literacy (Ormondroyd et al., 2014; Smart, 2010) have all been reported to impact family sharing and family members' decisions whether or not to pursue CGT. Specifically, the degree of emotional closeness or distance (Mesters et al., 2005; Ormondroyd et al., 2014), concerns over impact on family relationship (Stoffel et al., 2008), attitudes of family members (Wurtmann et al., 2018), and lack of understanding over meaning of results by patients and their relatives (McClaren et al., 2013; Stoffel et al., 2008) have been cited numerous times for their impacts on both disclosure and testing. In some studies, patients have reported confusion over who in the family was at risk for disease (Burns et al., 2016; Finlay et al., 2008), posing a barrier to accurate communication among relatives, and signaling an opportunity for healthcare providers to provide follow-up summaries with this information clearly outlined.

Importantly, low provider awareness is also described as a barrier to CGT uptake among relatives (Suttman et al., 2018; van El et al., 2018). As genetics moves out of specialty care and into general practice, it will be imperative that participating healthcare providers, irrespective of level of genetics training, have a solid understanding of genetics and CGT processes and workflows. Programs can develop tools to assist healthcare providers in supporting their patients, such as provider talking points and an electronic medical record built to more directly order targeted testing for family members (Haas et al., 2022), as well as materials or tools to directly share with patients such as genetic counseling notes or family letters. As an alternative, or in addition, health systems can also opt to grow a robust clinical genetics workforce to support elective genomic testing programs and subsequent CGT, an approach taken by Sanford Health (Blout Zawatsky et al., 2022). However, this may not be sustainable due to the ever-increasing demand for geneticist and genetic counselor expertise (Hoskovec et al., 2018; Jenkins et al., 2021). This will require building and sustaining partnerships with other healthcare providers like primary care physicians to carry out these programs (Blout Zawatsky et al., 2023). Furthermore, for family members seeking care outside of the original health system, workflows can be established to more directly connect them with genetics departments/laboratories and avenues to CGT within their own healthcare network.

Other studies have suggested additional, novel approaches to addressing some of the financial and logistic barriers to CGT, including offering no-charge testing (Courtney et al., 2019), online approaches with telephone genetic counseling (Caswell-Jin et al., 2019; Frey et al., 2020), mailed saliva-based genetic testing kits (Frey et al., 2020), and increased bioinformatic support to automate CGT processes (Haas et al., 2022). Geisinger, another major health system integrating elective genomic testing into primary care, has utilized a Family Sharing Tool and a Cascade chatbot to support family communication and CGT by allowing patients to electronically share

their genetic test results with their relatives via a chatbot (Campbell-Salome et al., 2021; Schmidlen et al., 2022). By employing such strategies and others at scale, elective genomic testing programs can alleviate reported barriers of CGT and optimize its facilitators.

Other findings of note include the pattern of information sharing within families. Patients with monogenic risk results in our study were more likely to report CGT in biological relatives than patients with carrier status results. This observed difference likely reflects an appreciation among patients that identification of monogenic risks can immediately impact the care of at-risk family members, whereas knowledge about carrier status may only impact reproductive choices (McClaren et al., 2013). We also observed that patients with carrier status findings shared their results frequently with their spouses/partners. This is an encouraging trend, as sharing carrier results with spouses has direct potential benefits for reproductive planning in those of childbearing age (McClaren et al., 2013). Interestingly, no patients with carrier status reported that their spouses or partners actually underwent CGT for the same disease predisposition for which they screened positive. While one might expect some uptake of CGT among spouses or partners based on carrier status findings (Ossa Gomez et al., 2022), it is important to note that the average age of carriers in this study was beyond childbearing age. We also observed instances where Sanford Chip findings appeared to prompt CGT in relatives, even when patients already knew about their variant. It is possible that re-disclosure of genetic risks provided an opportunity for genetic specialists to remind patients that their relatives had a high chance of having the same variants and risks for disease. In addition, we observed an instance where CGT motivated by Sanford Chip results was reported in a patient's relative with whom the results had not been shared, suggesting that results may have been shared with the at-risk relative by someone other than the patient. Such non-linear pathways to CGT highlight the importance of providing information about inheritance patterns and CGT that relatives, including spouses or partners who have not themselves been tested, can share with other family members to better ensure that at-risk relatives are informed of genetic risk factors that may impact their health.

4.1 | Limitations

We do not know the family sharing practices of the 15 patients with actionable predispositions who did not receive questions on family information sharing or CGT uptake because they did not finish the survey or because they reported not viewing or not remembering viewing their results. The fairly long period of time between results disclosure and survey completion (up to 4 years) raises risks of errors in recall about information sharing and CGT. Additionally, two patients had both monogenic risk and carrier status results and were reclassified as only having monogenic risk. Their responses were retained rather than being totally pulled because of potential bias, which poses a limitation to the analysis. Although patients were instructed in the survey to report sharing practices and CGT for biological relatives only, we cannot be certain that all reported family members are biologically related, nor the strength of relatedness (e.g., half siblings, half aunts/uncles). While analyses of sharing and CGT with a spouse/partner or child could be restricted to only those patients who reported having these family members, the survey did not assess whether patients had any other relatives; thus, all patients were included in those analyses, which could lead to an underestimate of the amount of sharing and CGT for those relatives.

Additionally, the survey did not evaluate the number of living relatives or family size of each patient. This depth would reveal more detail about the degree of sharing and CGT, as well as the number of relatives who were not disclosed to and the number of family members who did not undergo CGT. Similarly, the survey did not assess the number of family members per patient who underwent CGT for an actionable predisposition result, only the number of patients who had at least one relative undergo CGT. Future studies would benefit from eliciting this level of detail. The survey did not ask patients why they chose to share or not to share their actionable predisposition findings with their relatives. Likewise, it does not elicit information concerning relatives' reasons for or against CGT. Collecting this type of data would provide important context to the sharing and CGT data and would be beneficial to understanding barriers to and facilitators of family sharing and CGT. Moreover, while genetics visits were captured from medical records, it was not possible to discern whether visits were prompted by results from this study or by other referrals. Thus, we could not address the impact of speaking with a genetic counselor on sharing or CGT in relatives. Given the significant role genetic counselors often hold in supporting family sharing and CGT, these are important data to collect in future studies. Finally, primarily White patients were included in analyses, and the samples are small, limiting the power and generalizability of these data.

5 | CONCLUSION

Elective genomic testing has the potential to add significant value to precision medicine, particularly for at-risk family members through CGT. Findings from the Sanford Chip program in conjunction with learnings from other genetics research and clinical implementation projects can provide valuable insights for other elective genomic testing programs looking to optimize family communication and uptake of CGT. As healthcare professionals work toward integrating genomic testing and precision medicine into primary care, this work will require addressing these barriers through creative and strategic approaches that promote equitable access to CGT services.

AUTHOR CONTRIBUTIONS

Authors KDC and MRH confirm that they had full access to all the data in the study and take responsibility for the integrity of the data and the accuracy of the data analysis. All of the authors gave final approval of this version to be published and agree to be accountable

for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

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CONFLICT OF INTEREST STATEMENT

Robert C. Green has received compensation for advising the following companies: AIA, Allelica, Atria, Fabric, Genome Web, Genomic Life, Verily, and VinBigData; and is co-founder of Genome Medical and Nurture Genomics. Carrie L. Blout Zawatsky is compensated from Atria to coordinate and provide genomics education. Robert C. Green, Kurt D. Christensen, Emilie S. Zoltick, Madison R. Hickingbotham, Carrie L. Blout Zawatsky, and Sophia M. Adelson were supported by a research grant from Sanford Health. Catherine A. Hajek is an employee of Helix OpCo. Megan E. Bell, Dylan M. Platt, and Jennifer R. Leonhard declare that they have no conflicts of interest.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available from the corresponding author upon reasonable request.

ETHICS STATEMENT

Human Studies and Informed Consent: This study was approved by the Sanford Institutional Review Board and electronic consent was obtained from all participating patients.

Animal Studies: No non-human animal studies were carried out by the authors for this article.

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SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

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