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Experiences of adolescents and their parents after receiving adolescents' genomic screening results

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Abstract

There has been considerable debate over whether adolescents should have the opportunity to learn genetic information about adult-onset disease risk and carrier status without a clinical indication. Adolescents face increasing opportunities to learn more about such genetic risks through the return of secondary findings from clinical genomic testing, direct-to-consumer genetic testing, and research opportunities. However, little is known about the perspectives of adolescents who have received genomic screening results. We conducted separate qualitative interviews with 15 adolescents and their parents who enrolled in a research protocol where they decided which genomic screening results to receive for the adolescent for up to 32 conditions informed by 84

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AUTHOR CONTRIBUTIONS

Natasha Lillie contributed to study design, data collection, data analysis and interpretation, and drafting of the manuscript. Cynthia Prows and Michelle McGowan contributed to the conception, study design, data analysis and interpretation, and manuscript revision. Amy Blumling contributed to data analysis and interpretation and manuscript revision. Melanie Myers contributed to the conception, study design, data collection, data analysis and interpretation, and manuscript revision.

All authors confirm that they had full access to all the data in the study and take responsibility for the integrity of the data. Authors Natasha Lillie, Amy Blumling, and Melanie Myers take responsibility for the accuracy of the data analysis. All the authors gave final approval for 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.

CONFLICT OF INTEREST

Authors Natasha Lillie, Cynthia Prows, Michelle McGowan, Amy Blumling, and Melanie Myers declare that they have no conflicts of interest.

HUMAN STUDIES AND INFORMED CONSENT

This study was approved by Cincinnati Children's Hospital Medical Center (CCHMC) Institutional Review Board as part of a site-specific electronic Medical Records and Genomics (eMERGE) Network Phase III study (2016–3361) (Myers et al., 2020). Informed consent was obtained from all participants prior to being included in the study.

ANIMAL STUDIES

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

SUPPORTING INFORMATION

Additional supporting information may be found in the online version of the article at the publisher's website.

genes. The goal of these interviews was to explore the impact of adolescents learning genomic results without a clinical indication for screening. Of the participating dyads, four received positive results for a pathogenic/likely pathogenic (P/LP) variant for an autosomal dominant (AD) condition, five received carrier results for a heterozygous P/LP variant for an autosomal recessive (AR) condition, and six received negative results. An interpretive descriptive qualitative approach was used. Interview transcripts were coded using a guide developed by the study team based on themes that emerged from the interviews. Degree of recall and description of results, actionability, and emotional responses differed according to the types of results received. However, all participants were satisfied with their decision to learn results, and most did not report any lasting psychological harms. Participants adapted to genomic information about themselves, even after learning about unexpected increased risk for future health problems. Our findings support the position that, whenever possible, perspectives and wishes of adolescents should be strongly considered and respected in the decision-making process regarding genetic testing.

Keywords

adolescent; carrier testing; genetic testing; genomic screening; predictive genetic testing; psychosocial; return of results

1 | INTRODUCTION

As the availability of genomic testing increases, healthcare professionals must weigh the potential risks and benefits of adolescent participation in genomic screening for adult-onset conditions or carrier status in the absence of clinical indication. Historically, professional organizations recommended against genetic testing of adolescents unless a clear health benefit was present (ACMG, 1995) due to concern for potential adverse psychosocial impacts and to preserve the autonomy of adolescents until they could make independent choices as an adult (ACMG, 1995; Botkin et al., 2015; Ross et al., 2013). The American Society of Human Genetics (ASHG) and American College of Medical Genetics (ACMG) further recommend that adolescents wait until adulthood to have carrier screening, since information learned from carrier screening is not usually actionable until someone is planning to have children (Botkin et al., 2015; Ross et al., 2013). Despite considerable debate and recommendations against genomic screening for adult-onset conditions and carrier status, little empirical data exist regarding how adolescents are actually impacted by receiving these genomic results (Duncan & Delatycki, 2005).

Adolescence represents a unique stage in life when self-identity is developing and most individuals have yet to choose a career path, a long-term partner, or have children. Therefore, receiving positive genomic results may impact adolescents differently than adults, and a precautionary approach to allowing them to participate in genomic screening has prevailed. However, learning about a genetic predisposition may allow for preparation or prevention of future disease. Additionally, when adolescents learn their genetic status at a younger age, the information may more easily be integrated into their sense of self (Duncan & Delatycki, 2005).

The ACMG recommends that when a child has clinical genome or exome sequencing a set of secondary genes be analyzed and results returned in addition to the diagnostic results (Green et al., 2013; Kalia et al., 2017; Miller et al., 2021). Parents of children under 18 can opt in or out of this opportunistic genomic screening for their child, although, a separate committee within ACMG has encouraged parents and clinicians to involve the adolescent in the decision-making process and consider their preferences (Bush et al., 2018). It is recognized, however, that some adolescents may not possess the cognitive ability to think about the long-term impacts of receiving genomic test results, which may impact their ability to provide informed consent (Grootens-Wiegers et al., 2017).

Previous studies on the impact of adolescent genetic testing have largely focused on testing for a single condition due to family history, rather than genomic screening without clinical indication. Empirical studies on predictive testing in adolescents due to positive family history have not found a substantial risk for adverse effects on emotional well-being, self-perception, or relationships (Biesecker, 2016; Broadstock et al., 2000; Godino et al., 2016; Mand et al., 2013; Wade et al., 2010). In a systematic review of 11 qualitative studies that included between 8 and 44 participants, Godino et al. (2016) assessed the impact of predictive genetic testing on adolescents and young adults at-risk for adult-onset conditions, such as Huntington's disease, cancer predisposition syndromes, and familial cardiomyopathy. This review found that some participants with positive test results appreciated knowing what the future could hold and were able to make behavioral changes, apply the information to decisions about family and career planning, and enjoy life instead of facing constant worry about their genetic status. Although most did not experience significant adverse emotional impacts, some experienced depression and anxiety related to the risk of future disease or passing on the variant to children, the effect that the result would have on their career, and how they thought others may view them after sharing the result.

Studies on the impact of carrier screening results on adolescents have also failed to show significant risk for psychological harm (Botkin et al., 2015; Vears & Metcalfe, 2015; Wade et al., 2010). In a review of studies on how children aged six to nineteen years old were impacted by learning results of carrier testing due to family history or ancestry, Wade et al. (2010) found that those identified as carriers were satisfied with their decision to have testing and generally did not experience deleterious impacts on emotional state, self-perception, or social well-being. Carriers often reported that they would want to know the carrier status of their future partner for decisions about family planning (Wade et al., 2010).

The ways in which those who pursue genomic screening without clinical indication differ from those having single gene testing for a condition for which they are at known increased risk are unclear. However, there are increasing opportunities for adolescents to learn genomic information about disease risk and carrier status without a clinical indication through the return of secondary findings from clinical genomic testing, direct-to-consumer genetic testing opportunities, and research opportunities (Sabatello & Appelbaum, 2016), and parents and adolescents have demonstrated a desire for the option to learn this information (Fernandez et al., 2014; Hufnagel et al., 2016; Kleiderman et al., 2014; Levenseller et al., 2014; McGowan et al., 2018; Pervola et al., 2019; Sapp et al., 2014). To contribute to future evidence-based guidelines about adolescent genomic screening, we

explored the experiences of adolescents and their parents who chose to learn genomic screening results for multiple conditions for the adolescent in the context of a clinical research study.

2 | MATERIALS AND METHODS

This study was approved by the Cincinnati Children's Hospital Medical Center (CCHMC) Institutional Review Board (IRB) as part of a site-specific electronic Medical Records and Genomics (eMERGE) Network Phase III study (2016–3361).

A subset of adolescent/parent dyads who received genomic screening results for the adolescent through the eMERGE Network Phase III study at CCHMC were recruited to participate in separate qualitative interviews to explore their experiences of learning results and the impact it has had on their lives. Adolescents were aged 13–17 when they initially enrolled in the eMERGE Network Phase III study. Clinical indication for genetic testing was not an eligibility criterion for participation in the study. The adolescents made choices independently, and then jointly with their parent, about genomic results that they would like to learn. Only results upon which the adolescents and parents agreed were returned. The same adolescent/parent participants (dyads) participated in decision-making, result return, and qualitative interviews.

Our site-specific IRB-approved eMERGE III sequencing panel included 32 conditions informed by 84 genes (eMERGE Consortium, 2019), including the original set of genes recommended for return of results by ACMG (Green et al., 2013), and genes for AR conditions selected by some of the eMERGE sites (Myers et al., 2020; Pervola et al., 2019). One hundred forty-one adolescent/parent dyads received results that matched their choices. Adolescents with P/LP variants for AD conditions ($n = 5$) were given their results via phone and offered a follow-up appointment with a genetics professional. Both the adolescent and parent participated in the result call, except for one adolescent who gave permission to return the result by phone to the parent participant due to scheduling challenges. Adolescents who were heterozygous for AR conditions (carrier result, $n = 15$) or had negative results ($n = 121$) received results through an electronic patient portal.

2.1 | Participants

All adolescents who received heterozygous results for AD conditions or carrier results (positive results) and their parents were invited to participate in separate telephone interviews. For each adolescent receiving a positive result, an adolescent of the same sex and age who had received negative results was invited to participate in an interview. If an invitee with negative results did not respond, a second matched adolescent was invited. A total of 20 adolescent/parent dyads receiving negative results were invited to participate in an interview. Adolescents and parents were interviewed separately, and interviews were conducted until data saturation was reached. Participants were invited via email, followed by a text message after one week, and a final email reminder after one month. Recruitment took place from May through August 2020. Invitations were sent to parents and the adolescent if they were over 18. Adolescents and parents each received a $25 incentive for participation.

2.2 | Interviews

Semi-structured interview guides (one for adolescents and one for parents) were developed by the study team after a review of existing literature (Supplementary Material). The study team included members with expertise in qualitative research methods, genetic counseling, clinical genetics, and ethics. Interview questions addressed what participants remembered about their result and its health implications and their initial responses to the result. We also explored the impact of the results on emotional well-being, relationships, self-image, and future plans. Participants were asked what action(s) they had taken or planned to take in the future regarding the results, such as medical management, lifestyle changes, and sharing results with family members.

Verbal assent or consent was obtained at the beginning of each interview. All interviews were conducted by one member of the study team, with a second member participating in interviews of all participants with positive results and 60% of those with negative results. The lead interviewer was trained in conducting qualitative interviews by the study team and through pretesting the interview guide with a convenience sample. This interviewer was blinded to participants' genomic results and had not previously interacted with participants in earlier phases of the study. The researchers took notes during interviews which contributed to the development of codes and themes. Interviews ranged in length from 14 to 43 min for adolescents and from 12 to 65 min for parents. Interviews were audio-recorded and transcribed verbatim, and identifying information was removed from the transcripts.

2.3 | Data analysis

An interpretive descriptive approach was used for data analysis. Interpretive description 'is an applied qualitative approach to the uncovering of subjective and experiential knowledge relevant to the inquiry questions of the practice disciplines' (Thorne, 2016). Transcripts were coded using ATLAS.ti qualitative analysis software. A coding guide was developed with input from the study team based on themes that emerged from the interviews. Figure S1 shows the coding process that was used throughout data analysis to refine the coding guide (Saldaña, 2009). Two members of the study team coded each transcript independently using the coding guide and met to clarify codes and modify the code guide. All discrepancies between coders were discussed and consensus was reached.

3 | RESULTS

Our study sample included 15 adolescent/parent dyads. Characteristics of the adolescents are described in Table 1. All participating dyads identified as White. Average adolescent age at the time of the interview was 18. Time between the receipt of genomic test results and the interview ranged from 17 to 29 months (average 24 months). All parent participants were mothers and all mothers of adolescents with AD results reported having the same variant as their child. Only one mother was aware of her status before her child received testing through this study. No parents of children with a carrier result reported being tested for the variant found in their child before or after their child's result was returned.

Three primary themes emerged from interviews with adolescents and their parents regarding their experiences receiving genomic test results through this study: (a) Participant description and recall of result differed by result type, (b) perceived impact differed by result type, and (c) parents are stewards of their child's results.

3.1 | Theme 1: Participant description and recall of result differed by result type

Differences in how participants described their result and its implications and how they attributed meaning to their result were noted between those receiving AD, carrier, or negative results. Each group also differed in how well they remembered their results. Participants attributed a different level of importance to each result type. Those who found the result and its implications to be highly significant tended to remember their result more clearly and provided more detailed and accurate descriptions than those whose result did not raise concerns.

3.1.1 | AD result group—Both parents and adolescents with AD results used terminology similar to that used by medical professionals to describe the adolescent's result. Examples of phrases used by this group include: ‘a gene variant’, ‘a trait’, ‘a congenital heart condition’, ‘the cancer gene’, and ‘*BRCA* positive’. This group felt that the most meaningful aspect of learning their results was the symptoms or conditions that they could experience or had already experienced, how symptoms could be treated or prevented, and the possibility that other family members could have the same genetic variant. One adolescent did not remember the result very well but was able to describe the result implications:

> I had some sort of trait that would require me to come back later to make sure that I don’t have anything [along the lines of cancer] since it is in my genetics. But I didn’t have any active symptoms or anything like that
>
> (Male, 19, AD cancer predisposition)

Overall, the discussion of result implications was the most in-depth in this group.

3.1.2 | Carrier result group—Most of the adolescents who received carrier results, and their parents, described themselves or their child as being a carrier for a condition or a gene. Several described the results with phrases indicating that they did not feel the results were very significant such as: ‘something was abnormal’, ‘tests were negative’, ‘a small anomaly,’ ‘nothing really concerning’, and ‘not earth shattering’. This group felt that the most important aspects of the result were that the adolescent's future children could have a genetic condition, and that when the adolescent decides to have children, they can meet with a healthcare provider about reproductive options and risks.

Parents and adolescents in the carrier result group were more likely to state inaccuracies about their result than any other group. At least one member in four of the six dyads with carrier results made inaccurate comments about the result, such as listing incorrect symptoms of the condition, saying that this test identified them as a carrier for multiple conditions, or that being a carrier means you have or may develop the condition. In the remaining two dyads, at least one member did not remember any specific details of the result such as the name of the gene or condition, or the associated symptoms. Additionally, many

in the carrier result group mentioned they were unsure how to access their results in the electronic medical record (EMR).

3.1.3 | Negative result group—Adolescents who received negative results and their parents described the results with phrases such as ‘normal’, ‘no increased risk’, ‘nothing concerning’, ‘“no issues’, and ‘completely positive’. Dyads for whom the adolescent received negative results felt that the most important aspect of their results was that the adolescent was not genetically predisposed to any conditions tested. However, many recognized that they had a residual risk to develop the conditions. Two adolescents felt that their risks for developing the tested conditions were considerably lower after knowing their genetic results.

Those with negative results were less likely to remember the test results than any other group and many did not remember how to access their results.

> I don't remember getting back anything that – I mean, whatever we found out about him was kind of like not gonna make a huge difference. It didn’t make an impact
>
> (Parent of adolescent with negative result)

Several in this group did not remember if the results were positive for any conditions, stating that if there was anything abnormal, it was not memorable because it was unimportant. Others said they knew that their results were normal, but could not remember any conditions for which they tested negative.

3.2 | Theme 2: Perceived impact differed by result type

Participants described three main factors that summarized the overall impact of the test result including actionability of the result, emotional responses, and attitude toward test result. For each of these categories, similarities were found among participants within each result type group.

3.2.1 | AD result group—Most adolescents and parents in the AD group described actions they had taken or planned to take in the future regarding their test results. All parent participants had been tested, found that they have the same variant as their child, and had taken some medical action based on the result. All parents also reported sharing the results with other family members. In two families, at least one family member outside of the adolescent–parent dyad had testing. Participants with AD results experienced a wider variety and more emotional responses than participants with other result types. The strongest emotional reactions were expressed by adolescents and parents who received AD results that described current symptoms ($n = 4$) and/or that led to immediate medical intervention for themselves ($n = 4$).

Both adolescents with non-cancer-related AD results had experienced symptoms prior to receiving genomic test results without attributing them to a medical condition. These participants recalled feeling shocked at the initial diagnosis but had different reactions based on the prognosis of the condition. One adolescent and parent both had surgery following their test result to implant a device to monitor and regulate heart rhythm. Both described

lasting negative impacts on their mental health as a result of surgery, required lifestyle changes for the adolescent, and anxiety over future cardiac events. However, both expressed gratitude about learning the result and credited it with saving the adolescent's life because the device had prevented cardiac arrest on multiple occasions.

> ‘Emotionally, I probably am more affected than I am physically’… ‘If I’m being real, my quality of life is shit compared to what it was. I’ve lost a lot of relationships purely because my health isn’t good, and they don’t want to be associated with that’…. ‘And then, I’ve had people leave because of stress from the surgeries or my mental health not doing good’… ‘I don’t know if it’s been good or bad. It’s been good and bad, but I can’t tell which one outweighs the other’. When asked if he would still choose to learn genetic results if given the choice, he said ‘Absolutely, I’d be dead if I didn’t’.
>
> (Male, 17, AD)

> The amount of stress and anxiety he must be feeling all the time from it is overwhelming to me. He's doing pretty well, but I still worry. It’s a lot to deal with. I feel guilty for spreading it, giving him the variant. It’s hard’…. ‘So, I just try and remember all the time how grateful I am that we didn’t find out by someone dying, because most of the people in the support group that I participate in, they found out because they lost [someone]. So, we are so lucky.
>
> (Parent of adolescent with AD result)

In the other dyad, the adolescent, his mother, and another sibling had all experienced symptoms. The mother had been extensively evaluated as an adult, but doctors had not identified a cause prior to genetic testing. She was elated and relieved that her son got a result that explained her own symptoms.

> And then [child] gets his results, and it's like, oh my goodness! This explains everything! So, it was awesome to be able to go to the doctors when the test results came back for me and to be able to go to them and say, okay, it's not MS or ALS, this is my genetic test result. And then, knowing that this is genetic, being able to have family members tested down the line.
>
> (Parent of adolescent with AD result)

Her adolescent son was also grateful but did not attribute the same significance to the results as his mother, as his symptoms were milder:

> ‘Before the results I was a little anxious, but then when I heard it, I'm like… I don't wanna say relieved is the exact word, but a little more understanding. Sort of like, oh, okay, yeah, I understand this. If [symptom] happens again, I can explain what it is and so that way if people see it and worry, I can be like, oh, it's fine, it happens every now and then’… ‘It's just a little mutation. It's nothing too big’.
>
> (Male, 18, AD)

Adolescents who learned about increased cancer risk reported that they planned to start cancer screening at ages recommended by their doctor, and one had changed their medication to reduce cancer risks. One adolescent who had chronic health concerns reported

that when she received the genetic result, she considered it in the context of her other unrelated health concerns, including a previous life-threatening issue, and it did not seem as significant in comparison. She described being initially overwhelmed by the genetic test result because she had not heard of it before but was eventually glad to know the information because of management options available to her.

> It's allowed our family to think more deeply about our health choices and how we're living our life, and allowing us to get better imaging, and just be able to care for our bodies better.
>
> (Female, 16, AD cancer predisposition)

A second adolescent was aware prior to this study that his mother had a known cancer predisposition variant that was identified after a cancer diagnosis. This adolescent expressed minimal emotional response to the result because prior to enrolling in the study, he felt he would need testing in the future for it anyway. His mom reported feeling guilty for passing the variant to her son, but was glad to know about his risk so she can help establish a plan for him to get screened in the future.

> 'I cried. I was sad. Yeah, I was sad that I had passed it down. He was more optimistic. He was like' 'It's okay, Mom'. 'And then after I thought about it, I was better. I was like, it's better to know than not to know'.
>
> (Parent, AD cancer predisposition)

Two adolescents reported changes to their future plans regarding having children, stating that they may not want biological children because they do not want to pass their genetic variant on or because of other health conditions in addition to the test result.

> With how I've seen it affect me, but also my parents looking at me like, "Wow, we really gave our kid this," I don't want to feel like that. I don't want anyone else to go through this. It's always been my dream to have a nuclear family, and now that's shifted. I used to really, really want kids, and now I'm just like, I don't wanna pass it on. Unless they somehow find a way to alter our babies and make it so they don't have a variant of the [gene], then I don't want them.
>
> (Male, 17, AD)

> I have pretty much decided that I'm not going to have biological children personally, but it depends if I find someone. I mean it's hard to say that because I'm only 16, so if I find someone that I feel like really wants biological children… but I think fostering and adopting children are more my future child plans.
>
> (Female, 16, AD cancer predisposition)

These two also discussed changes to their career plans. One reported wanting to become a doctor to improve treatments for their condition, and another intended to have a low stress job to minimize cancer risks.

3.2.2 | Carrier result group—Adolescents with carrier results and their parents reported that they felt the results will be more relevant and actionable when the adolescent is considering having children. None had taken any action at the time of the interview, aside

from two parents who informed their child's pediatrician and/or other family members about their child's result. No family members of any adolescents with carrier results had been tested. Two adolescents said that they will speak to a health professional when ready to have children to learn about reproductive risks and options, as well as how the condition could impact their child. One adolescent said that her cystic fibrosis transmembrane conductance regulator (CFTR) carrier result encouraged her to 'be safer when it comes to intercourse' and make smart decisions about choosing a partner because she is at risk of having a child with a serious condition.

> I think I would definitely want genetic testing for my partner and then, make decisions from there if they are also a carrier. I feel it would kind of be unfair to not go through a process of looking at the chances to have a child with [condition] because it's definitely a very serious disease and they could have to spend a lot of years in the hospital, and it's very scary. Then maybe if there's a really high chance that our kid would have [condition], think about that before we decide to have kids. And then if maybe that's adopting or having some sort of, I guess, donor. I don't really know.
>
> (Female, 19, carrier result)

> 'I'm not really thinking about my future as far as having kids right now. Maybe down the road if it comes to that I will, but I didn't really have any plan in the first place. So, not really a huge change because of my age, but I think maybe once it comes down to that, maybe I'll have to talk to a doctor about that, just to see what the risks are'…. ' I don't think it would necessarily make me not have children, but it would just be something that I would maybe talk to my doctor about before having kids. I'm still going to have kids no matter what, but what is this going to mean if they have it? How can we prepare to make life easier for them if they do have it?'… 'I think that with modern medicine, I'm not too worried about it, but I did – I did find it interesting to learn about'.
>
> (Female, 17, carrier result)

A common response to learning carrier results was that it could be worse, comparatively speaking. Participants felt that the risks for passing on the condition were not that high, or if they did have a child with the condition, it would be manageable. The adolescent who was found to be heterozygous for an AR and AD condition did not mention his carrier results, and instead only spoke about his AD result, which he clearly felt was more impactful. None of the carrier result group members mentioned lasting emotional impacts from learning results. Most expressed relief that the only result they received was a carrier result. A few expressed initial sadness and surprise, but after learning more about the condition and options to review the results with a professional when planning a pregnancy, those who initially felt sadness found the implications of the results to be manageable.

> When I actually found them out, I was super grateful that being a carrier for cystic fibrosis was the only thing that I tested positive for and was a carrier for. Because, I mean, it's still something really deadly and a scary disease, but it wasn't in my eyes the worst thing that could have happened.

(Female, 19, carrier result)

One parent expressed relief because her child was negative for everything except the carrier result, so they would not have to worry about cancers that have affected previous family members:

> This has just had such a tremendous benefit for both of us because I think it gave him a lot of peace of mind. It gave me some peace of mind as well because we didn't have to worry about watching out for the different cancers that have come up in my family in the past.

(Parent of adolescent with carrier result)

Many expressed that the results brought them peace of mind because they did not receive results that placed themselves of their child at increased risk for developing any conditions on the panel.

3.2.3 | Negative result group—No participants in the negative result group had taken any action based on their results or reported plans to take action in the future. Only one parent said they informed their child's pediatrician of the negative result. One adolescent said the process of being in the study inspired them to live a healthier life because it made them think about their long-term health.

Dyads receiving negative results for the adolescent expressed relief, peace of mind, or no emotional reaction. As a result of adolescent negative results, some parents expressed that they or their other children likely also have a good chance of being negative.

> It's just the peace of mind for me, to know that since she is negative that the likelihood of her siblings being negative is – is pretty good.

(Parent of adolescent with negative result)

Many adolescents in this group felt the results gave them peace of mind because they were not at increased risk for any of the conditions tested.

3.2.4 | Overall attitudes—All participants were satisfied with their decision to learn the adolescent's genomic results.

> I think it's good to know as much as you can about your own genetics so that you can prepare for the future even if you are just in adolescence.

(Male, 19, AD cancer predisposition)

They felt that all adolescents should have the option to learn genetic information about themselves but that the decision to have testing should depend on the individual. For example, several parents suggested adolescents should only have genetic testing if the adolescent is mature enough and ready for the information.

> I guess now we have power. Knowledge is power. So, now we have an opportunity to make choices about the diagnosis, so to speak. It's not really a diagnosis, but the genetic results. We have a choice now, so now you have the power to change an

> outcome, and so for me, it was a decision I would do again. I don't have any regrets about it.
>
> (Parent of adolescent with carrier result)

Adolescents and parents commonly stated that knowledge is power when it comes to learning genetic information, even if it is not actionable.

3.3 | Theme 3: Parents are stewards of their child's results

Interviews revealed that parents often served as a resource when their child had questions about their genomic screening result. Adolescents depended on their parent to help them interpret the result and put it in context of their family history.

> I was definitely familiar with [gene] based on my family history. So, [child] did not know what it was, but I was well aware. And so, to me it wasn't necessarily shocking and to her she didn't necessarily understand exactly what that meant yet.
>
> (Parent of adolescent with AD cancer predisposition)

Additionally, parents were most often the ones to share test results and information about the condition with the adolescent's primary care provider (PCP) or other at-risk family members. Some adolescents admitted that they did not know if their PCP or other relatives had been told about the results, but that their parent probably knew or shared the result. Several parents also indicated they would hold onto the results and remind their child about them when they were more relevant or actionable.

> Next time she goes to her primary physician, we'll probably have her take this in. I'll keep a copy. And then, you know, once she gets older, decides to have children, or thinking about that, then I would want her OB to see it.
>
> (Parent of adolescent with carrier results)

Adolescents depended on their parent to manage their EMR and to know or find out how to access the results. This was problematic for parents of adolescents who had turned 18 since receiving the results, because some of them lost access to their child's records after they turned 18. Several adolescents stated that they never saw a copy of their results as their parents verbally relayed the information to them, particularly in the carrier and negative result groups.

4 | DISCUSSION

While many have posed theoretical impacts of predictive genetic testing and carrier screening in adolescents, this is the first study to explore the actual experiences of adolescents who have received genomic screening results for multiple conditions without a clinical indication for testing. Results from our interviews with adolescents and their parents who had received genomic test results 17–29 months prior suggest that degree of recall and description of results, actionability, and emotional responses varied by result type received. However, all participants were satisfied with learning results, regardless of result type received. Our findings suggest that adolescents and their parents were grateful

to receive genomic results for conditions that they chose to learn, even if they identified an increased risk for unexpected future health problems.

As noted, all adolescents expressed satisfaction with their decision to learn genetic results, two thirds of whom had turned 18 since choosing to receive results. The cognitive capacity and emotional maturity of adolescents of the same age may differ (Duncan & Delatycki, 2005; Grootens-Wiegers et al., 2017), and it is unreasonable to expect that all adolescents will reach maturity to make informed choices at a specific age or at the point at which they are legally considered adults. Duncan and Delatycki (2005) urged clinicians to seriously consider the possibility that adolescents may reach this maturity much younger than 18. Adolescents in our study who underwent testing as young as 13 years old reported satisfaction with their decisions about learning and receiving genomic screening results 17–29 months later. For adolescents who may already be experiencing symptoms of a condition or are considering having children, genomic information could be especially beneficial to know before reaching the age of majority to understand the cause of their symptoms or make informed reproductive decisions. Our findings align with those of other studies that have found that adolescents who have had clinically indicated predictive or carrier testing do not later regret the decision to have testing (Mand et al., 2013; Wehbe et al., 2009). It is important to note that in our study, testing decisions were made after a joint discussion between the adolescent, their parent, and a study team member, where the perceived risks and benefits of learning genetic information were discussed in depth (Pervola et al., 2019). This joint decision-making process supports ACMG's recent recommendations for a dialog between adolescents, parents, and clinicians that strongly considers the adolescent's wishes when deciding to learn about secondary findings from clinical genomic testing (Busch et al., 2018).

Recommendations against genomic screening of adolescents for adult-onset conditions and carrier status sought to protect adolescents from adverse psychosocial impacts of testing and to preserve their decision-making autonomy until they can make independent and informed choices as an adult (ACMG, 1995; Botkin et al., 2015; Ross et al., 2013). The 'child's right to an open future' rationale coined by Feinberg outlines the right to keep options open for when a child reaches adulthood (Feinberg, 1980). This concept has been applied to pediatric predictive genetic testing decisions because once a parent or child learns genetic or genomic test results, the child no longer has the option not to know that information and is therefore stripped of their right to choose as an adult (Davis, 1997). However, conceptual harms and professional society recommendations were established without robust empirical evidence of adverse outcomes in adolescents who have received genetic test results (Duncan & Delatycki, 2005). Most adolescents whom we interviewed reported no lasting negative psychosocial impacts due to learning the genomic screening results 17–29 months prior. Similar findings have been reported by others who examined the impact of receiving predictive genetic testing results based on adolescent family history (Godino et al., 2016; Wade et al., 2010).

While most adolescents having testing without a clinical indication will likely receive negative results or not be significantly impacted by the results, some will receive unexpected or life-altering results which may be immediately relevant and actionable during

adolescence. The possible need for medical intervention when the adolescent is a minor highlights the importance of keeping parents and legal guardians engaged in the decision-making process, as they are the legal decision makers for the adolescent's medical care until the adolescent reaches the age of majority. Our findings also suggest the important role parents play as a resource for their children, providing support and helping them understand their results in the context of their family history. Previous research has shown that many adolescents prefer their parents to be present when receiving information about their genetic condition, but some would rather prefer their parents not to be present for discussions about recurrence risks and prognosis (Pichini et al., 2015). Our findings underscore the importance for clinicians and researchers to work with adolescents to understand their personal wishes regarding parental involvement, within legal limits.

One important finding of this study is the difference in recall and descriptions of the implications of the result. Those with carrier results were most likely to make inaccurate comments about the result or forget the result implications and several participants with negative results forgot their results. Botkin et al. (2015) expressed concerns about the ability of result recipients to remember genetic results accurately over long periods of time, and although our participants could access their results through an EMR, several expressed that they did not know where to find the results. Participants in our study who received negative results often reported their risk was not zero but could not remember specific conditions for which they were tested. The potential need for follow-up after returning negative results was reflected by the parent who felt her child would not have to worry about cancers that had affected past family members. Similar false reassurance should be anticipated when sequencing is used for clinical purposes and negative secondary results are returned. The differences in what the carrier and negative groups remember about their result compared to the AD group may be attributed to how the results were returned or the immediate actionability of the results. Those with AD results received results from a genetic counselor by phone and were then offered a follow-up visit with a genetic counselor or a specialist, while those with carrier and negative results received results through an electronic patient portal. Although the implications and limitations of results were described in the electronic health record (EHR) report, participants who were not personally contacted about their results may have been less inclined to think the results were important. Several parents suggested that it would have been helpful to receive a phone call no matter what the results were to ensure understanding of the results and give an opportunity for participants to ask questions. Although all participants were given the option to talk with a genetic counselor after receiving results, none reached out directly by their own initiative. This finding suggests that future research ought to assess different modes of returning carrier and negative genomic screening results to participants to assess their understanding and recall of results. Although not specifically addressing recall, interviews with and surveys of adult participants receiving negative genomics screening results by postal mail or online also found that while participants valued learning their genomic screening results, some reported lack of understanding or misunderstanding of their results (Butterfield et al., 2019; Cheema et al., 2021; Hoell et al., 2020; Stuttgen et al., 2020).

4.1 | Practice implications

Our findings have important implications for genetic counselors who may work with adolescents and their families in clinical or research settings where adolescents have the opportunity to choose to learn genomic results, such as through the return of secondary findings from genome or exome sequencing. While allowing seemingly healthy adolescents to choose to receive genomic screening results has raised questions about the long-term psychosocial impacts of such results, our findings show that in the context of a clinical research study, adolescents and their parents were grateful to have the information and were able to adapt to the results, even after learning about increased risk for unexpected future health problems. These findings are consistent with those of other studies involving adolescents who have chosen to receive predictive genetic testing or carrier screening results that have not found evidence for lasting adverse psychosocial impacts or regrets about the decision to have testing. This study adds evidence toward refuting historical reluctance to involve adolescents in the decision-making process regarding genetic testing. Our findings support the position that, whenever possible, perspectives and wishes of adolescents should be strongly considered and respected.

4.2 | Study limitations

Limitations include our small sample size, particularly within each result type group, as well as enrollment through one academic medical center. Additionally, all participating adolescents identified as White, which closely reflects the composition of those receiving positive results in our site-specific study, but does not reflect the composition of the population as a whole or of the region in which the study took place. It is also noted that our interviews took place with adolescent/parent dyads who were willing to discuss their experiences, and there may have been a selection bias for those who had better memory of their experience or different responses than those who did not respond to invitations to participate. However, there have been many calls for research into the experiences of adolescents who have received genomic screening results, and we have been able to capture the perspectives of those who have received results for a variety of conditions. While we were able to hear from adolescents who have known their results for 17–29 months, those with cancer predisposition syndromes or carrier results were not yet at an age where they found their results to be actionable, and it would be helpful to know if results inform decisions or actions later in life.

Our qualitative methods allowed us to capture the experiences of adolescents who received genomic screening results and how they viewed them in the context of their own family histories and life circumstances. Additional studies are needed to understand the impact of learning genomic screening results on participants from diverse backgrounds, as previous research suggests adolescents who are Black may make different decisions than adolescents who are White (Myers et al., 2020). It may be useful for future studies to utilize quantitative methods, such as validated psychological assessments, to assess the long-term impact of learning results.

5 | CONCLUSIONS

While degree of recall and description of results, actionability, and emotional responses varied by result type received, adolescents and parents did not express regret after receipt of genomic results for the adolescent. Our findings highlight the importance of keeping parents engaged throughout the testing process, as they are important for helping adolescents navigate access to the results and follow-up actions. Overall, adolescents in this study remained satisfied with their decisions to learn genomic information about themselves after receiving results. In addition, they adapted to these results even after learning about increased risk for unexpected future health problems. This study adds evidence toward refuting historical reluctance to involve adolescents in the decision-making process regarding genetic testing, supporting the position that perspectives and wishes of adolescents should be strongly considered and respected when possible.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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DATA AVAILABILITY STATEMENT

The research data for this study are not shared publicly.

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What is known about this topic

Adolescents face increasing opportunities to learn genomic information about adult-onset disease risk and carrier status from the return of secondary findings from clinical genomic testing, direct-to-consumer genetic testing, and research opportunities. Historically, professional organizations recommended against genetic testing of adolescents unless a clear health benefit was present to avoid potential adverse psychosocial impacts and to preserve the autonomy of adolescents until they could make independent choices as an adult. Despite considerable debate and recommendations against adolescent genomic screening, little is known about the perspectives of adolescents who have chosen to receive genomic screening results.

What this paper adds to the topic

While allowing seemingly healthy adolescents to choose to receive genomic screening results has raised questions about the long-term psychosocial impacts of such results, our findings show that in the context of a clinical research study, adolescents and their parents were grateful to have the information and were able to adapt to the genomic information even after learning about unexpected increased risk for future health problems. While degree of recall and description of results, actionability, and emotional responses varied by genomic result type received, all participants were satisfied with their decision to learn results. Findings from this study add strength to the case against historical reluctance to involve adolescents in the decision-making process regarding genetic testing, supporting the position that, whenever possible, perspectives and wishes of adolescents should be strongly considered and respected.

TABLE 1

Adolescent demographics

Characteristics	***n***
Sex	
Female	8
Male	7
Result received (gene with heterozygous P/LP variant[a])	
CACNA1A (AD)	1
BRCA1 (AD)	1
SCN5A (AD)	1
CHEK2 (AD)	1
CFTR (AR)	2
SERPINA1 (AR)	4
No P/LP identified (negative)	6
Age	
At consent for testing	Average: 15, range: 13–17
At result return	Average: 16, range: 13–17
At interview	Average: 18, range 16–20

Note: Among the 20 adolescents who received positive results, all but one identified as White, 14 were female, and the average age at the time of result return was 15.

Abbreviations: AD, autosomal dominant; AR, autosomal recessive; LP, likely pathogenic; P, pathogenic.

[a]One participant received heterozygous results for an AD and AR condition.