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# The beliefs, motivations, and expectations of parents who have enrolled their children in a genetic biorepository

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**Purpose:** Little is known about parental attitudes toward return of individual research results (IRRs) in pediatric genomic research. The aim of this study was to understand the views of the parents who enrolled their children in a genomic repository in which IRRs will be returned.

**Methods:** We conducted focus groups with parents of children with developmental disorders enrolled in the Gene Partnership (GP), a genomic research repository that offers to return IRRs, to learn about their understanding of the GP, motivations for enrolling their children, and expectations regarding the return of IRRs.

**Results:** Parents hoped to receive IRRs that would help them better understand their children's condition(s). They understood that this outcome was unlikely, but hoped that their children's participation in the GP would contribute to scientific knowledge. Most parents wanted to receive all IRRs about their child, even for diseases that were severe and untreatable, citing reasons of personal utility. Parents preferred electronic delivery of the results and wanted to designate their preferences regarding what information they would receive.

**Conclusions:** It is important for researchers to understand participant expectations in enrolling in a research repository that offers to disclose children's IRRs in order to effectively communicate the implications to parents during the consenting process.

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**Key Words:** biorepository research; individual research results; parent perspectives; pediatric biobank; pediatric genetic research; returning research results

## INTRODUCTION

As advanced genomic research studies have begun to reveal the significant influence of genes on health and disease, a contentious debate has emerged about whether or not researchers should return individual research results (IRRs), defined as research findings specific to a study participant,[1,2] to subjects. Some have cited the return of IRRs as an "ethical imperative" of researchers,[3] and public polling suggests that offering IRRs serves as a strong incentive for large numbers of people to enroll in genetic biobanks.[4,5] Others argue that offering IRRs may persuade people who otherwise would not have participated in genetic research to do so because of a misguided hope of personal gain when there is in fact no guarantee of direct benefit from research.[6] In addition, there is a concern that the return of IRRs blurs the distinction between research and clinical care and may elicit a therapeutic or diagnostic misconception among participants.[7]

Opinions about the involvement of children in large-cohort genetic studies are mixed, and both public opinion studies and professional editorials raise a number of ethical concerns,[8–11] which center around the child's transition from partial to full autonomy. Issues include the need to obtain meaningful assent from minors, and full informed consent when the child turns 18 years of age, in order to ensure their continued participation in the study. An additional concern is how to balance the disclosure of results to parents, who make decisions on behalf of the child, with respect for the child's future autonomy in controlling their own research results; this may be a particular challenge in the case of research results with reproductive implications for the child and the parent. On the other hand, studies of return of research results in children and adolescents with cancer suggest that parents and children have a strong desire to receive research results.[12–14] To date, however, most of the information collected on attitudes about returning research results in pediatric populations is based on surveys or interviews with participants who are not actively enrolled in research studies in which results are returned.

Despite these concerns, the debate has expanded to include not only the question of whether or not to return the IRRs, but, if results are returned, what results to return and how to return them. The current suggested guidelines use criteria for return of IRRs that focus on analytic validity, clinical validity,

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actionability, and severity of the outcome.[15] However, some argue that the personal meaning of genomic information to participants,[16–18] participant preferences, and personal utility[19–22] should play an important role in return of research results. The "multidimensional results reporting" model proposed by Kohane and Taylor[16] incorporates participant preferences, "communicability"[16] of the result (how likely it is that the message about a result will be understood), and the significance of the result in the decision-making process for return of individual genomic information.

Taking the latter approach to the return of research results in genomic studies, members of our group, in the publication of Kohane et al.,[23] proposed the informed cohort as a model for returning genomic research results to participants that accounts for their changing involvement over time and allows participants to take an active role in research.[23] The informed cohort model takes advantage of the advances in information technology by facilitating ongoing communication with research participants through a web-based personally controlled health record (PCHR). Through the PCHR, researchers can notify participants about IRRs according to individual preferences regarding what types of results they would most like to receive. A key feature of the informed cohort is the informed cohort oversight board (ICOB), a group similar to the institutional review board but with additional expertise in risk communication, genetics, and genetic counseling. The ICOB is responsible for determining what information is worthy of communication, how best to ethically and responsibly communicate it without confusing or overwhelming participants, and how to help participants set preferences regarding what research results they would like to receive.

Aspects of the informed cohort, including the ICOB, have been adopted by other groups.[15,24] Our group at Children's Hospital Boston (CHB) has implemented the informed cohort model in a pediatric setting by establishing the Gene Partnership (GP), the first longitudinal genetic repository that enables research of the genetic and environmental influences on childhood health and disease and offers to return IRRs under the guidance of an ICOB. Through the GP database, de-identified genotypic data, family history information, and data uploaded from the electronic medical record will be available to researchers. In order to receive communications from the GP, participants create a PCHR account and designate their preferences regarding what types of research results to receive. If there is an IRR that is consistent with the participants' preferences, they receive a message through the PCHR asking them to call in and speak with a genetic counselor to learn about the IRR and discuss the finding.

The consent process for participants is thorough, and key elements discussed include the following:

1. GP data will be available to investigators to carry out research on a variety of health conditions.
2. Participants can choose whether or not to receive IRRs and in the future will have the opportunity to decide what types of results to receive by setting preferences.
3. Participants should not necessarily expect to receive IRRs about their child related to the condition(s) for which their child is followed at CHB.
4. Participants may receive IRRs about their child related to conditions other than those for which their child is followed at CHB;
5. Results from the study are not clinical, are not recorded in the medical record, and are not disclosed by the GP to the primary care physician; and
6. IRRs for children regarding adult-onset conditions for which there is no treatment or prevention will not be returned to parents, which is in accordance with current clinical genetic testing guidelines[25–27] and respects the child's right to decide, when they become fully autonomous, whether they do or do not want such results.[28]

Children aged 7 years and older who are of normal cognitive ability are assented to the study and are contacted again at the age of 13 years to re-assent to ensure that they are comfortable participating in the study and with their parent(s) receiving research results on their behalf. At 18 years of age, participants are contacted and must undergo the informed consent process to remain in the study. If a participant does not re-consent at age 18 years, their data are removed from the GP database and are not used in future studies.

In order to determine whether participants who enrolled in the GP accurately understood the project as discussed during the consenting process, and to gain insight into the views and preferences of parents whose children are enrolled in the repository, we conducted focus groups with parents who had enrolled their children in the GP. The project was in a pilot phase at the time of our study and families had been recruited into the GP through either the Developmental Medicine Center (DMC) or the Genetics Clinic at CHB. As a result, most of the parents who participated in our focus group study had children diagnosed with complex developmental and/or genetic disorders. We asked these parents to discuss their understanding of the GP, motivations for enrolling their children, and their preferences and expectations with regard to receiving their children's IRRs. We were interested in determining whether parents had received proper informed consent for the GP and in providing feedback to the ICOB as it develops policies for the disclosure of children's IRRs.

## MATERIALS AND METHODS

### Recruitment

English-speaking parents who had a child enrolled in the GP were eligible for the study. All families enrolled in the GP as of May 2010 ($N = 122$) received a contact letter inviting one parent per family to participate and received a follow-up telephone call to assess interest and availability. At the time of study, 98% of the families enrolled in the GP were recruited through the DMC clinic and the remaining 2% through the Genetics Clinic. A total of 19 parents participated in one of the three focus groups.

The study was approved by the CHB institutional review board, and informed consent was obtained from all participants.

### Focus group discussions

We asked parents to discuss (i) their understanding and concerns about the GP project; (ii) their motivations for enrolling their child in the GP; and (iii) their expectations and preferences with regard to receiving research results. We began the focus groups by asking parents to describe the GP in their own words. After this discussion, the moderators provided a brief overview of the GP to ensure that all participants remembered the key details and differentiated the return of IRRs (research findings specific to a study participant) from the return of aggregate (general and not participant-specific) results. It should be noted that because parents can enroll both their children and themselves in the GP, a parent may receive IRRs about their child when their child's sample is used in a study and IRRs about themselves when their sample is included in a study. In the focus groups, we asked parents to focus on their feelings with regard to receiving IRRs about their child. The same two moderators (E.D.H. and S.I.Z.) led the three focus group discussions using a moderator guide (**Table 1**); both have previous training and experience in moderating focus groups.

The focus groups were held in June, July, and August of 2010 at the CHB satellite location in Waltham, MA. Each session consisted of 4–9 participants and lasted 1–2 h. There was a note-taker for each session (J.G.A.), and the discussions were audio-recorded for accurate data collection. Before the start of the focus group, demographics were collected on all participants. After the discussions, participants were compensated with a $50 Visa gift card.

### Analysis

Audio recordings of the focus groups were transcribed by an external agency, and members of the project staff cross-checked transcripts with audio files and notes from the sessions for accuracy. Two research assistants (E.D.H. and J.G.A.) reviewed the transcripts independently, generated separate de novo lists of common themes that surfaced in the focus group sessions (using questions in the moderator guide as framework), and then reconciled the lists to create a catalog of the most prevalent themes. Comments in the transcripts were assigned codes to correspond with one of the theme categories. Coded comments were then organized within each category by the speaker to ensure that no theme was overrepresented simply because one participant had repeated the same idea multiple times in one session. This allowed the study staff to identify the most prevalent themes established in the focus groups.

## RESULTS

### Participants

Participants were primarily Caucasian (95%) and female (79%) and ranged in age from 33 to 58 years (**Table 2**). The highest level of education attained was graduation from a 2- or 4-year college for eight participants and completion of postcollege courses or a graduate degree for nine participants. Previous experience with research was reported by 9 participants, and 11 had prior experience with genetic testing. All participants had at least one child followed in the DMC, four parents had more than one child followed in the DMC, and several of these children had multiple diagnoses. Parents reported the following diagnoses for their children: 10 children had an autism spectrum disorder (ASD), 6 children had attention deficit hyperactivity disorder/attention deficit disorder, 4 children had a genetic syndrome, and 3 children had a seizure disorder. One child each had a metabolic disorder, oppositional defiant disorder, and an unknown diagnosis. A variety of diagnoses were represented in each focus group, and each session contained at

**Table 1** Moderator guide

| Outline of focus group prompts |
|---|
| What do you remember about GP? What, in your words, is the purpose of the project? |
| Why did you enroll your child or family in the project? |
| Did you have any fears or did anything cause you to hesitate when you enrolled? |
| What do you expect as "results," in your own words? |
| Consider that you could get many different research results back about your child from the GP database—which of these results would you want to be messaged about? Why? |
| • Results for which there is no known treatment or prevention |
| • Severe or fatal results |
| • Uncertain or preliminary results |
| • Low relative-risk results |
| • Results about adult-onset conditions |
| How do you want to receive results? What should be included and who should results be delivered to? |

GP, Gene Partnership.

**Table 2** Participant demographics

| | Group A | Group B | Group C | Total (%)[a] |
|---|---|---|---|---|
| *n* | 7 | 8 | 4 | 19 (100) |
| **Age (years)** | | | | |
| Mean | 43.7 | 46.6 | 47.7 | 46.0 |
| Range | 37–58 | 33–58 | 36–57 | 33–58 |
| **Gender** | | | | |
| Male | 0 | 3 | 1 | 4 (21) |
| Female | 7 | 5 | 3 | 15 (79) |
| **Race** | | | | |
| White | 7 | 7 | 3 | 17 (89) |
| Black | 0 | 1 | 0 | 1 (5) |
| Declined | 0 | 0 | 1 | 1 (5) |
| **Highest level of education** | | | | |
| High school or equivalent | 0 | 0 | 0 | 0 |
| Some college/ vocational school | 0 | 0 | 1 | 1 (5) |
| College degree | 2 | 4 | 2 | 8 (42) |
| Postgraduate courses/degree | 5 | 4 | 0 | 9 (47) |
| Declined | 0 | 0 | 1 | 1 (5) |
| Previous research participation | 5 | 2 | 2 | 9 (47) |
| Previous genetic testing (participant or child) | 4 | 5 | 2 | 11 (58) |
| Diagnosed with genetic disorder (participant or child) | 2 | 1 | 2 | 5 (26) |

[a]Percentages may not total 100% due to rounding errors.

least one parent of a child with ASD and one parent of a child with a genetic syndrome. Children ranged in age from 4 to 17 years old at the time of the focus groups, and the ages were distributed as follows: six children aged 4–6 years, seven children aged 8–11 years, five children aged 12–15 years, and two children aged 16–17 years.

### Understanding of the GP and concerns about the project

When asked what they understood about the GP, participants accurately identified the GP as a research repository designed to facilitate research of genetic contributions to childhood diseases. They also understood the broad nature of the repository: one parent pointed out, "it's not going to be so tailor-made that I'm going to find out everything about our children" (parent of a child with a genetic syndrome).

When asked whether they had experienced any concerns with regard to providing their child's genetic material for research, some participants noted transient concerns about confidentiality and loss of insurance, which they immediately dismissed because they trusted CHB to keep their children's genetic information private and secure:

> Had it been some small hospital or some smaller group I never would've done it. But I think with the backing of an institution like Children's, the fear crossed my mind but I got over it really quickly (parent of a child with a genetic syndrome).

### Motives for enrolling and requesting research results

When parents were asked what motivated them to enroll in the GP, two themes arose. The key factor that participants pointed to was hope that they would receive IRRs that would provide information about their child's health condition(s), which would eliminate confusion about the etiology and progress of the child's diagnosis, help them prepare for the future, and possibly relieve feelings of guilt. The second motivating factor for participants was the potential that aggregate findings from research studies using the GP database would lead to scientific advances in understanding childhood disease that would benefit society.

### Personal benefits of receiving the IRRs

Sixteen of the 19 participants expressed feelings of confusion, isolation, and/or frustration regarding their children's diagnoses and enrolled in the GP hoping that the study might help them better understand their children's medical conditions. "We keep getting different answers," one parent shared, "it's like we're on a rollercoaster" (parent of child with genetic syndrome). One participant reported that her daughters' conditions were so rare that the girls were "writing their own medical books" (parent of two children with genetic syndromes), and another participant commented, "I've got 17 different specialists in Children's that we see for one thing or another [...] and nobody there has the big picture" (parent of child with unknown disorder). Parents felt that their children's current diagnoses did not adequately describe the children's complex medical issues. Even parents who had clear diagnoses for their children expressed feelings of isolation and a desire for more information: "When you're looking for answers, you're willing to go anywhere […] because you're in it all by yourself" (parent of child with a genetic syndrome). Parents hoped that the GP would yield answers that would help them understand their child's condition, allowing them to "put a piece together" to solve the "puzzle" of their child's health (parent of child with genetic syndrome).

Over three-quarters of the parents expressed hope that through the GP they would get IRRs about their child's condition(s) that would help them forecast the progression of the child's health, manage expectations, and prepare for the future.

> We're looking for answers, as well as maybe to help us with expectations of where our kids may be 10 years from now, from every aspect of the word. Health-wise, neurologic related, in any way. Just to know the full picture of perhaps—give us our roadmap (parent of child with genetic syndrome).

Some parents hoped that the GP would provide information that would help their child obtain services, such as additional

supervision at school. "There's a system you have to game at some level to get the right things for your kids," one father said, "and this kind of information could help us be able to do that in a better way" (parent of child with unknown disorder).

Several parents said that receiving an IRR that identified a genetic cause for the child's condition would confirm that the condition was not the result of something that the parent had or had not done, thus eliminating their sense of guilt. In one mother's words, results from the GP might answer the question, "did you eat too much tuna fish [when pregnant], or really do you have somebody in the family that has a neurological condition?" (parent of child with genetic syndrome). Several participants shared personal anecdotes about family mysteries that had been solved by genetic testing. One participant talked about the guilt that her mother endured after bearing a child with a seizure disorder and the subsequent relief that she felt upon finding a genetic cause for his condition:

> My mother thought, "I should've asked for a C-section," and she carried that around for 30 years. And blamed herself—really blamed herself for it. So when we found out he had that [genetic] deletion, giving her that information, I think, was a huge weight lifted off her shoulders (parent of child with ASD).

### Altruistic motives for enrolling

Despite a strong drive for answers related to their child's health condition, parents maintained an overall awareness that their family would not necessarily receive personal benefit from research studies conducted using the GP database. They demonstrated an understanding that the project may not provide the results they would like, but instead may yield indeterminate and nondiagnostic findings and acknowledged the long-term nature of the GP, and the potential to never receive any results. Nonetheless, they claimed that the greater potential benefit to society was strong enough of an incentive to participate.

> I don't expect anything, but I'll be retired, if not dead, I think, before true results [are returned]. I say, cure? Ah, I don't think it's going to help us any. But if something comes out of it that can help people further down the road, further the medical field of study, hey, I'm all for that, that's good (parent of child with ASD).

Participants seemed to understand that any IRR they might receive would not be clinical or diagnostic in nature and therefore should not be treated as more than research findings. "You're just telling us what you found, but it's not a diagnosis," one mother articulated (parent of child with ASD), suggesting that, at least in this cohort of participants, therapeutic misconception was not an issue. In addition, parents understood that IRR could indicate an increased risk of a disease and not that the participant has a disease. Another parent added, "You could find something that never happens," (parent of child with unknown disorder), highlighting that participation may also yield insignificant findings.

Participants instead expressed hope that research from the GP would serve as a foundation for scientific research about children's health, feeling that medical information available on the internet did not represent "real scientific research" (parent of child with ASD). One parent characterized involvement in the GP as "scientifically philanthropic"—enrolling not only for one's own family, but for "the greater good of humanity," and others agreed, finding solace in the fact that "something as simple as spitting into a cup may have huge ramifications for the people in the future" (parent of child with ADD).

### Participant preferences for receiving an IRR

When asked which types of research results they would like to receive, almost every participant reported that they would like to receive all research findings and expressed the sentiment that knowing any information was better than knowing none: "We don't want to have our head in the sand if there's something that you can do" (parents of child with genetic syndrome). Although they wanted all research results, many also felt that parents in general should be given a choice of what results to receive and suggested a system that would allow parents to "set filters" about what information they would like to be notified about—"That way you can get the results that you're comfortable with, and take whatever action you want to take to start the discussion" (parent of child with ASD).

When asked to consider receiving an IRR about a severe or life-threatening condition in their child, almost all parents wanted to be informed because of the personal utility of the results: "when you live in a situation that is really unsure and it's very tenuous, like any kind of information is welcome" (parent of child with genetic syndrome). When moderators qualified this by asking if parents would like to learn about severe results that were also untreatable, parents felt that, even in the absence of actionability, such results would help them "[make] the best life you possibly can for your child with what's left" (parent of a child with ASD). Only two parents said they would not want information about nontreatable or nonpreventable conditions in their children. One parent stated that knowing such a result would be like living with a "cloud overhead" and would infringe on her children's right to autonomy:

> I'm not sure that I would want to know that, and carry the weight of that burden through their lives, until they were ready to maybe make the decision to know or not know […] I'm not saying I never want to know. I'm sort of saying I don't want to make that choice for them when they're this age that they should know or not know (parent of child with ASD).

When asked about receiving IRRs that were not "well established" (new findings not yet supported in the literature), most parents reported that they would like to receive the result, concurring, "we are all reasonably adept at dealing with uncertainty"

(parent of child with an unknown disorder). Parents pointed to their experience in the DMC, where they felt that the diagnoses their children were given were not as distinct or clear as those assigned in other clinical departments, as evidence that they are well equipped to receive ambiguous research results. In reflecting on the potential to learn uncertain results from the GP, one parent noted, "We actually already have that. It was one of those—it's a [genetic] duplication and we've never seen it before, but we'll call you if we find anything. So I love that. I think it's great. I want to know" (parent of child with ASD). Parents in all groups agreed that they would approve the disclosure of such results as long as they were provided with context: "If it's something new that you just find out and it's probably going to take you three or four years to sort it out […] then say that" (parent of child with unknown disorder).

Parents across all three focus groups felt that they should receive contextual information along with the IRRs: "You have to give a little bit of information about the science to people" noted one parent, "because sometimes information can be misunderstood" (parent of child with ASD). They suggested that families be guided to resources that explain research findings in detail, links to relevant literature if available, and recommendations for next steps including experts who could help them interpret results or support groups. In one parents' words, "It would be negligent to give someone information of potential horrific diagnosis in the future without giving them some ability to handle it with some expert" (parent of child with ASD).

Most participants also preferred electronic delivery of results, which would allow them to "access [results] on their own accord" (parent of child with genetic syndrome), giving them time to digest the information. In response to one parent's comment that she would like her children's physician to deliver the results, several parents pointed out that a primary care physician may lack the expertise needed to fully interpret genetic research findings, particularly novel ones, and that they feared results could become "lost in the shuffle" of "busy and overbooked doctors" (parent of child with genetic syndrome).

## DISCUSSION

Studies suggest that return of the IRRs is a motivating factor for participation in genomic research[4] and that parents and children have a strong desire to receive research results from pediatric genetic studies.[12–14] Ours is the first study of parents who have enrolled their children in a genetic research repository, the GP, that is not disease specific and thus returns IRRs on a broad range of conditions investigated by researchers using the repository. Participants in our study conveyed an overall comprehension of the purpose of the GP and conveyed few fears about enrolling their children in genomic research.

Similar to studies about the return of research results in adults,[4,5] our participants reported that the potential to receive an IRR specific to their children was a strong incentive to enroll their children in genetic biorepository research. A major finding was that participants hoped to receive IRRs that would explain the genetic cause of their children's underlying conditions, and that the utility of such findings was largely personal. They hoped that the IRRs would clarify their child's diagnosis, alleviate feelings of guilt about the cause of the condition, allow them to better prepare for the future, and help their child obtain additional services. The desire to understand their child's health may be a particularly strong sentiment in our study because many of the parents had experienced uncertainty in the clinical setting with regard to their child's diagnosis. In addition, a number of parents had a child with ASD, and the drive for answers may be particularly great in this population.[29] It was notable that, despite this strong desire for IRRs that would explain the child's condition, parents understood that it was unlikely that they would get answers to their specific questions. They were motivated to enroll a child in the GP in hopes that their family's participation would lead to general research results that would contribute to global scientific knowledge and help children in the future.

In general, our study population wanted to learn all the IRRs about their children, regardless of severity, actionability, or validity of the information. Comments from the focus groups indicated that parents viewed a personal utility in receiving IRR's for their children, even regarding severe or untreatable conditions, and felt that the knowledge would help them to manage their expectations and provide their children with the best life possible. This finding emphasizes that the benefits of receiving results are personal, fully dependent on the views, values, and environment of each participant.[16]

Of note, at least one parent was concerned that learning IRR's related to severe, nontreatable conditions would infringe on a child's autonomy, as well as burden the parent. This serves to remind us how crucial it is to both respect a child's autonomy in the return of research results[9] and to acknowledge the potential negative effects that disclosure could have on parents. The disparity in views between parents who wanted to receive all research results and parents who were hesitant about receiving certain types of results points to the potential value of creating tools, such as the PCHR proposed by the GP,[23] through which participants can define their preferences and learn only about those results that they are comfortable knowing. In fact, several participants suggested disclosure policies that would allow them to choose which results to receive, and nearly all participants supported electronic delivery of results, which would allow them to access results on their own accord. This desire for electronic delivery of results is important for the GP to consider as the study moves forward because the current method for returning results is contacting participants through the PCHR to inform them that a result is available and asking parents to call in and speak with a genetic counselor to discuss the results. Participants also unanimously reported that they expect to receive contextual information along with their child's IRRs, particularly those of unclear significance, to help them interpret the results and direct them to resources for gathering more information.

### Limitations

There are several limitations to our study. Our sample represented a small, select group of parents who had children with

complex developmental and genetic disorders. Their unique experience with diagnoses that are not always clearly defined may account for their frustration, confusion, and subsequent appeal for as much information as possible about their children's health. Parents of children with ASD may be particularly susceptible to such feelings[29] because the genetic influences in the majority of affected children are still unclear. Additionally, the motivations for parents of children with different conditions may differ substantially. Organizing the focus groups to have parents whose children have similar conditions might have led to clearer discussions, but this was not feasible in our study given the small number of families available to participate. Additionally, our population consisted of primarily female (79%), highly educated parents (95% had a college degree or higher level of education) and was not racially diverse (89% Caucasian). Future studies to address these issues could involve a wider scope of parents, include parents of children with a greater variety of medical issues, and potentially divide parents into more homogenous groups

Because the enrollment in the GP and in the subsequent focus-group study were both voluntary endeavors, our participants also represent an especially proactive population. Their willingness to volunteer for a focus group, as well as their past experience in research studies (over half reported previous participation in research), is evidence of these parents' involved, hands-on approach to learning about their children's health and may account for their seemingly firm grasp of the GP study concept as well as their stated interest in receiving all types of research results. Parents who take a less active role in research may have a more difficult time understanding the purpose of a research repository, whereas those who are less involved in their children's healthcare or lack strong motives for enrolling may have a different set of expectations of the project. Nonetheless, any parent who chooses to enroll a child in the GP has, in doing so, inherently demonstrated a level of interest in research and may subsequently have some expectation that results will arrive with contextual information.

Finally, although one parent expressed concern about learning IRRs that would infringe on the child's autonomy, we did not fully explore issues related to the parent–child dynamic inherent in disclosure of research results in a pediatric setting, such as how participants might balance their feeling that receiving their child's IRRs would be in the child's best interest with their child's increasing autonomy over time and right to an open future. Additional studies will be needed to address the views of children, especially adolescents, about research participation and sharing of their genomic information with their parents. The approach we are undertaking is to convene focus groups of adolescent participants themselves who could provide insight into additional subtleties and considerations of disclosing results in a pediatric population.

### Conclusions

We think that we have taken an important first look at a population whose beliefs and opinions about child participation in genetic research will be increasingly important to consider as researchers begin to disclose results from pediatric genomic research. Participants demonstrated a strong desire to receive all types of research results and for control of data that are relevant to their family. Their comments suggest that learning their children's IRRs, regardless of clinical utility, would make them feel more in control of their children's health and better prepared for the future. These perceived benefits of learning the IRRs about one's child including those about late-onset disorders, which extend beyond the recommendations to return only clinically actionable results,[15] suggest that researchers should consider a result disclosure policy that more fully acknowledges personal utility of findings.

Underlying our study findings, there is also a valuable message about the importance of the informed consent process. The informed consent discussion provided for enrollment into the GP appears to have provided the parents with realistic expectations of the research project. We did not see evidence of a significant therapeutic misconception, or the blurring of research results with clinical care,[7] and overall parents accepted that, although they would like to learn the IRRs about their children's specific conditions, they are unlikely to directly benefit from the GP participation. Furthermore, parents' desire to receive their children's IRRs in an electronic format illustrates a shift toward a more contemporary practice through which patients and study participants may like to learn personal genetic information in the future.

Overall, our study provides insight into the experiences and motives of families who enroll in a genomic research repository and highlights parents' desire for control over information about their children's health and the importance of effective communication between researchers and participants.

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## DISCLOSURE

The authors declare no conflict of interest.

## REFERENCES

1. Bookman EB, Langehorne AA, Eckfeldt JH, et al.; NHLBI Working Group. Reporting genetic results in research studies: summary and recommendations of an NHLBI working group. *Am J Med Genet* A 2006;140: 1033–1040.
2. Knoppers BM, Joly Y, Simard J, Durocher F. The emergence of an ethical duty to disclose genetic research results: international perspectives. *Eur J Hum Genet* 2006;14:1170–1178.
3. Fernandez CV, Kodish E, Weijer C. Informing study participants of research results: an ethical imperative. *IRB* 2003;25:12–19.
4. Kaufman D, Murphy J, Scott J, Hudson K. Subjects matter: a survey of public opinions about a large genetic cohort study. *Genet Med* 2008; 10:831–839.
5. Murphy J, Scott J, Kaufman D, Geller G, LeRoy L, Hudson K. Public expectations for return of results from large-cohort genetic research. *Am J Bioeth* 2008;8:36–43.

6. Meltzer LA. Undesirable implications of disclosing individual genetic results to research participants. *Am J Bioeth* 2006;6:28–30; author reply W10.
7. Clayton EW, Ross LF. Implications of disclosing individual results of clinical research. *JAMA* 2006;295:37; author reply 37–38.
8. Burke W, Diekema DS. Ethical issues arising from the participation of children in genetic research. *J Pediatr* 2006;149(1 Suppl):S34–S38.
9. Kaufman D, Geller G, Leroy L, Murphy J, Scott J, Hudson K. Ethical implications of including children in a large biobank for genetic-epidemiologic research: a qualitative study of public opinion. *Am J Med Genet C Semin Med Genet* 2008;148C:31–39.
10. Jenkins MM, Rasmussen SA, Moore CA, Honein MA. Ethical issues raised by incorporation of genetics into the National Birth Defects Prevention Study. *Am J Med Genet C Semin Med Genet* 2008;148C:40–46.
11. Ries NM, LeGrandeur J, Caulfield T. Handling ethical, legal and social issues in birth cohort studies involving genetic research: responses from studies in six countries. *BMC Med Ethics* 2010;11:4.
12. Fernandez CV, Gao J, Strahlendorf C, et al. Providing research results to participants: attitudes and needs of adolescents and parents of children with cancer. *J Clin Oncol* 2009;27:878–883.
13. Fernandez CV, Santor D, Weijer C, et al. The return of research results to participants: pilot questionnaire of adolescents and parents of children with cancer. *Pediatr Blood Cancer* 2007;48:441–446.
14. Fernandez CV, Taweel S, Kodish ED, Weijer C. Disclosure of research results to research participants: A pilot study of the needs and attitudes of adolescents and parents. *Paediatr Child Health* 2005;10:332–334.
15. Fabsitz RR, McGuire A, Sharp RR, et al.; National Heart, Lung, and Blood Institute working group. Ethical and practical guidelines for reporting genetic research results to study participants: updated guidelines from a National Heart, Lung, and Blood Institute working group. *Circ Cardiovasc Genet* 2010;3:574–580.
16. Kohane IS, Taylor PL. Multidimensional results reporting to participants in genomic studies: getting it right. *Sci Transl Med* 2010;2:37cm19.
17. Ravitsky V, Wilfond BS. Disclosing individual genetic results to research participants. *Am J Bioeth* 2006;6:8–17.
18. Rothstein MA. Tiered disclosure options promote the autonomy and well-being of research subjects. *Am J Bioeth* 2006;6:20–21; author reply W10–W22.
19. Foster MW, Mulvihill JJ, Sharp RR. Evaluating the utility of personal genomic information. *Genet Med* 2009;11:570–574.
20. Grosse SD, Kalman L, Khoury MJ. Evaluation of the validity and utility of genetic testing for rare diseases. *Adv Exp Med Biol* 2010;686:115–131.
21. Grosse SD, McBride CM, Evans JP, Khoury MJ. Personal utility and genomic information: look before you leap. *Genet Med* 2009;11:575–576.
22. Khoury MJ, McBride CM, Schully SD, et al.; Centers for Disease Control and Prevention. The Scientific Foundation for personal genomics: recommendations from a National Institutes of Health-Centers for Disease Control and Prevention multidisciplinary workshop. *Genet Med* 2009;11:559–567.
23. Kohane IS, Mandl KD, Taylor PL, Holm IA, Nigrin DJ, Kunkel LM. Medicine. Reestablishing the researcher-patient compact. *Science* 2007;316:836–837.
24. Stack CB, Gharani N, Gordon ES, Schmidlen T, Christman MF, Keller MA. Genetic risk estimation in the Coriell Personalized Medicine Collaborative. *Genet Med* 2011;13:131–139.
25. Points to consider: ethical, legal, and psychosocial implications of genetic testing in children and adolescents. American Society of Human Genetics Board of Directors, American College of Medical Genetics Board of Directors. *Am J Hum Genet* 1995;57:1233–1241.
26. Report on the Genetic Testing of Children 2010. Report of a working party of the British Society for Human Genetics. Presented at Wolfson College, Oxford, UK, March 2008. http://www.bshg.org.uk/GTOC_Booklet_Final_new.pdf.
27. Committee on B. Ethical issues with genetic testing in pediatrics. *Pediatrics* 2001;107:1451–1455.
28. Hens K, Nys H, Cassiman JJ, Dierickx K. The return of individual research findings in paediatric genetic research. *J Med Ethics* 2011;37:179–183.
29. Miller FA, Hayeems RZ, Bytautas JP. What is a meaningful result? Disclosing the results of genomic research in autism to research participants. *Eur J Hum Genet* 2010;18:867–871.