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Translating Genomic Testing Results for Pediatric Critical Care: Opportunities for Genetic Counselors

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Abstract

Genomic sequencing (GS), such as whole genome and exome sequencing, is rapidly being integrated into pediatric critical care settings. Results are being used to make high impact decisions including declarations of futility, withdrawal of care, and rationing of scarce resources. In this qualitative study, we conducted interviews with clinicians involved in the care of critically ill children with congenital heart disease (CHD) to investigate their views on implementation of GS into clinical practice. Interviews were transcribed and inductively analyzed for major themes using grounded theory and thematic analysis. Three major themes emerged surrounding the use of genomic information in the high-stakes, time pressured decision making that characterizes clinical care of critically ill children with CHD: (1) that clinicians felt they did not have sufficient training to accurately assess genetic results despite pressure to incorporate results into clinical decisions; (2), that they desire knowledge support from genetic specialists, such as genetic counselors, who both understand the critical care context and are available within the time constraints of critical care clinical pressures; and (3), that clinicians feel a pressing need for increased genetics education to be able to safely and appropriately incorporate GS results into clinical decisions Our data suggest that genetics specialists may need a stronger presence in the pediatric critical care setting.

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AUTHOR CONTRIBUTIONS

DC designed the study. DC conducted the interviews and primary coding of the transcripts. ND, SL, and DC analyzed the data. ND and DC drafted the manuscript. All authors commented on and reviewed the final manuscript.

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Natalie Deuitch, Sandra Soo-Jin Lee & Danton Char declare that they have no conflicts of interest.

Keywords

genome sequencing (GS); critical care; congenital heart disease (CDH); education; ethics; genomics; genetic counselors; genetics education; whole genome sequencing (WGS)

INTRODUCTION

Rapid GS has been piloted and is being adopted in pediatric and neonatal critical care settings (Clark et al., 2019; Farnaes et al., 2018; Petrikin et al., 2015; Priest et al., 2014; Stavropoulos et al., 2016). Conditions seen in this setting can be diagnostically complex and often co-occur with other diseases. Furthermore, many of these conditions warrant high-risk treatments and early and precise diagnosis is critical. Rapid clinical trio exome sequencing of infants with suspected genetic diseases has been reported to yield up to a 50% diagnostic rate, and can have significant impacts on medical management (Meng et al., 2017). Advances in sequencing technologies have drastically decreased both cost and processing time. Analysis that previously took months can now occur in a matter of days (Clark et al., 2019; Miller et al., 2015). The potential for rapid and cost-efficient diagnostics is leading many institutions to develop in-house GS clinical services with the expectation of an exponential increase in their use over the next few years (Kapil et al., 2019; Willig et al., 2015).

Critical care genomic sequencing is especially applicable for infants with congenital heart diseases (CHD). CHD is the most common type of birth defect leading to critical illness in the United States (Hoffman & Kaplan, 2002; Reller et al., 2008) and is the leading cause of birth defect-associated illness and death (Yang et al., 2006). Given the complexity of disease and the highly invasive and costly procedures often required for children with CHD, such as organ transplants, the care of this patient population often involves difficult clinical decisions for both clinicians and their family members (Morell et al., 2012).

Previous studies have assessed the technical aspects of integrating genetic sequencing to critical care settings such as turn-around times, cost effectiveness, and medical actionability of results (Clark et al., 2019; Kapil et al., 2019; Stark et al., 2018). They have looked at parental experiences with GS in NICU settings and have stressed the need for genetic counseling for families considering such testing (Ayres et al., 2019). However, implementation of GS in critical care settings may also have profound impacts on the clinicians who care for these children.

Results from GS can be complicated and critical care physicians receive limited training in how to interpret them. Detection of variants of uncertain significance (VUSs), incidental findings, and results with implications outside of the ordering physician’s area of expertise can make GS results difficult to interpret. If acted upon incorrectly these results can have profound medical implications for the patient. Currently, the number of practicing geneticists and genetic counselors are inadequate to address results for all patients who could receive GS – particularly in the setting of the time-sensitive decisions that are often involved in critical care (Hoskovec et al., 2018; Ormond et al., 2010). With such time pressures, the burden of interpreting and contextualizing GS results often falls on bedside

ICU clinicians despite their having limited knowledge and understanding of genetics and genetic testing (Baars et al., 2005; Char et al., 2018; Marzuillo C et al., 2013).

In critical care settings clinicians and families must continually assess whether the invasive therapies offered are proving useful or are prolonging needless suffering, weighing the uncertainties of treatment success against the pain and suffering inflicted by the treatment choice (Boss et al., 2012). GS is being implemented in this population and has the potential to impact on how these difficult care decisions are made (Char et al., 2018; Priest et al., 2014; Stavropoulos et al., 2016; Willig et al., 2015). Implementation studies of GS in pediatric critical care have shown that transition to palliative care was more often identified as the clinical outcome of genomic sequencing than other actions such as changes to medication, procedures or counseling (Char et al., 2018; Petrikin et al., 2015; Willig et al., 2015). Furthermore, rapid GS can be used to support decisions to proceed or not with surgical interventions in neonates (Miller et al., 2015; Petrikin et al., 2015). Making such grueling and critical decisions from GS data certainly elicits ethical concerns (Char et al., 2018).

To better understand the experience of pediatric critical care clinicians with early implementation of GS information to clinical care, and the potential ethical and practical challenges that could come with its implementation, we interviewed 35 clinicians caring for critically ill children with CHD about their experiences and thoughts about this new technology. This paper describes the themes that emerged from these interviews regarding their role in understanding and utilizing GS results, as well as their thoughts on what support is needed for successful implementation.

METHODS

We used a qualitative approach, interviewing physicians, nurse practitioners and physician assistants involved in the care of critically ill children with CHD at a high-volume pediatric heart center and other care centers within the same system. Interviews were collected between March 2016 and May 2018.

Interviewees were initially approached via telephone or email. Potential interviewees were stratified based on characteristics such as clinical specialty and seniority to encompass the diversity of clinicians caring for critically ill children with heart disease at the heart center. One-on-one interviews were conducted in-person to provide an intimate, private context for discussing sensitive topics (such as personal decision-making) and because this approach is well suited for exploratory research attempting to find a range of perspectives (Christensen et al., 1992; Feveile et al., 2007; Lemaire & Wallace, 2010; Olson, et al., 2013; Poulin & Edu, 2010; Ullström et al., 2014; Yoon et al., 2010). The study was approved by the IRB of the Stanford University School of Medicine.

A semi-structured interview guide was developed and was piloted with 5 clinicians/participants. Questions included current and hypothetical ethical considerations of GS implementation (Supplementary Table 1: Questions). Interviews were conducted either in person or over the telephone and were audio-recorded and transcribed. Transcripts were

uploaded into the qualitative analysis software Dedoose, (www.dedoose.com) and interviews were analyzed incorporating grounded theory (Charmaz, 2014; Clarke, 2005; Corbin & Strauss, 2015). Codes were generated inductively through a collaborative reading and analysis of a subset of interviews (DC,SL,MC) and then finalized through successive iterations into categories and codes. At least one primary (DC) and one secondary coder (AI) independently coded each transcript. Differences were reconciled through consensus coding. The team collaboratively reviewed each code and discussed interpretation of themes in a series of consultations (Ryan & Bernard, 2003). Emerging themes were identified, described and discussed by the research group. Interviews continued until thematic saturation was achieved.

RESULTS

Study participants included 35 clinicians (Table 1: Demographics). 100% of those invited to participate completed the interview. Interviews lasted approximately 40 minutes, but ranged from 20–60 minutes. Overall, participants expressed a need for support using GS results in caring for critically ill children. Thematic analysis identified three areas of perceived gaps in support: 1) lack of clinician training and ability to interpret genetic results 2) a call for more genetics specialist support in critical care settings; and 3) clinicians desire ongoing education about GS to keep pace with implementation.

1) Lack of clinician training and ability to interpret genetic results

The majority of clinicians saw utility and potential for GS to be used in the high-stakes decision-making that occurs in critical care settings. Several participants described using GS sequencing to inform treatment decisions, like recommending a particular surgery, but also used it to make decisions about withholding treatment or transitioning patients to palliative or comfort care.

> “If a child is stuck on a ventilator and can never come off and is going to die of a horrible respiratory disease within a few weeks, no parents want to put their child through that amount of suffering, so GS can make a lot of hard decisions easier. But we have to learn ourselves and be able to teach others how. Hopefully it won’t make that many decisions harder.” (P34, Neonatologist)

However, many also expressed significant concern about interpreting genetic results and using them to make such decisions. These clinicians recognized that genetic findings had significance outside of their realm of clinical practice, and could have life-altering implications for their patients. This concern was exacerbated by the fact that many interviewees felt doubtful about their own knowledge of genetics and genomics, especially given the rapid pace of advancement in the field.

> “I trained, started my medical school six years after chromosomes were first done, so I go back! And a lot has been learned since that time, only some of which I know about.” (P6, Neonatologist)

Several clinicians clearly recognized that their knowledge of genomics was less than sufficient to interpret GS, especially to the level required to make critical clinical decisions.

The majority of participants had received only limited formal training in genetics. This was especially true for more senior physicians who had trained before many key advances in genetics and genetic testing.

> "When I went to medical school, I mean Mendelian genetics was it. A monk taught us genetics!" (P1, Neonatologist)

While more senior clinicians often couched their responses in jokes about age, this concern about inadequate genetics training was also present among junior clinicians. Many commented about the limited training that they had received in medical school, even more recently.

> "I definitely feel woefully uninformed or inadequately informed about this topic … I don't know, it's hard to keep up with the current knowledge base that we're expected to know." (P3, Intensivist)

Additionally, a few interviewees commented that they were concerned that medical education was moving away from rigorous formal study of the genome, making interpreting GS results even harder for clinicians.

> "Here in the medical school we're cutting back dramatically on all the preclinical courses. There's a proposal to whittle it down to a year and a half rather than two years, and it's not even a year and a half of full time because half the time is spent in practice of medicine. So they're not even getting the basis to be able to understand this stuff [GS] in medical school." (P8, Cardiologist/Geneticist)

2) Call for more genetics specialist support in critical care settings

Given concerns about interpreting GS results, all participants articulated a strong need for interpretation and decision support with GS use. Clinicians who had worked with genetic counselors valued them immensely when they were available.

> —"I rely incredibly heavily on our genetics counselor. I rely on her like 100% and she's been an incredible asset to our division. I don't know how people in other institutions who don't have that kind of resource are doing this [GS] and doing this kind of logically and rationally because there's no way to figure out what's important and what's not important. It's very helpful to have somebody translate." (P1, Cardiologist)

While many clinicians noted that they were limited in their ability to stay abreast of genetic information, several commented that a geneticist or genetic counselor could fill in that gap and play a role in interpreting GS results. For these clinicians the breadth of information in GS results made it difficult for them to even know what was important to consider in making clinical decisions. A number of interviewees felt it was unrealistic to expect them to master the depth of knowledge required interpret results in addition to their already difficult task of caring for critically ill children.

> "To have a genetic counselor that can say, these are the things that are important, this is what it means rather than having to have us understand all of the test results and what that means and what the ramifications are, or what the ramifications

> aren't, because I think then that's where it kind of opens the kind of can of worms where you just don't have the time or complete understanding to be able to answer all of that." (P4, Intensivist)

One participant likened interpretation of GS results to that of CTs scans in critical care decisions. When it comes to CT scans a radiologist has expertise in scan interpretation and works with the clinician to develop a plan for patient care. They felt the same could be true for results from GS. A physician may not have the necessary training to interpret the results and may need to collaborate with a genetics specialist.

> "Why we have experts over-reading CT scans is partly to protect us from not having to understand every single technology that we use, and so the same thing would go here [with GS]. You would expect an expert to read the genetic test result and for you, as a physician, to be responsible for reading the approved test results… But it's somewhat of a new world where we get information on so many different diseases" (P12, Cardiologist/Geneticist)

Several clinicians recognized that, unlike radiology where a radiologist could provide a comprehensive interpretation of the diagnostic imaging through over-reading, with GS there is not a clear model of knowledge support for the burden of contextualizing GS findings across so many potential clinical implications. Consequently, these clinicians desired genetic counseling support when making clinical decisions from GS results. However, many felt that support was often hard to find.

> "Our genetics program is not, at least clinically, a service that has been very accessible to us." (P17, Intensivist)

Many commented on the potential for problems should support not be available, especially given the rapid integration of GS. Clinicians were uncertain of who would provide needed GS support, since geneticists and genetic counselors were not often accessible or included in making acute or high-stakes decisions.

3) Desire for ongoing education about GS

While most participants saw that genetic counselors would serve a valuable role in providing the clinical support needed to use GS in critical care, the majority also recognized that there is a professional responsibility to understand the fundamentals of GS. This was especially pertinent given the impact that GS results could have on their specific patient population.

> "If our patient population is going to be a good target population for this type of approach – and certainly congenital heart disease is an excellent target – then I think we need to better understand what the issues are, what the technology is, what the limitations of the technology's results are. It should be entering into med school curricula, it should be part of residency training and fellowship training curricula, as we go forward, and then ought to be part of continuing medical education for practitioners who are already out and about." (P30, Cardiac Intensivist)

One participant noted that while a genetic counselor may be able to provide guidance on GS, it is also imperative that the bedside clinician be able to integrate various genetic risks with other clinical information to develop a comprehensive care plan.

> "It's the nine blind men and the elephant, they're looking at that little part of the patient that might be abnormal, might be affected by a genetic aberration, or a genetic combination, and I don't know that we're looking at those things together." (P7, Cardiologist)

Participants repeatedly commented on a desire for further education in genetics. For some the need was more urgent, as genetic results were already being used in their practice. Others felt that there had not been the clinical pressure to learn about GS yet, however, they could foresee it becoming more important in the future as the technology becomes more ubiquitous.

> I certainly think it should be embedded in all of our ongoing medical education as the technology… But, you know, in my work, I don't have many times where I think I wish I really understood this more because it doesn't…current state, you know, it isn't affecting what I'm doing day to day, but again, I don't think it's far off that it will be. (P25, intensivist)

While most participants recognized that they would not have the same training as a genetic specialist, they did feel that a basic understanding of GS would help them improve patient care. Participants identified a number of competencies that they thought would be valuable to understand. These included: how to interpret a result, applications and limitations of the technology, how GS works, as well as ethical issues surrounding GS.

> "I know a little bit about genetics, I'm not a geneticist, I'm a cardiologist, but I have some…and I have a lot of patients who have cardiogenetic disorders and so I know some of the basics. But WGS/WES, pros and cons and differences in targeted gene testing, I think this is the thing that a lot of cardiologist and general practitioners know very little about, so I would be very in favor of more education on this." (P28, Cardiologist)

Clinicians clearly recognized that the rapid implementation of genomic sequencing was outpacing their ability to learn about genetics on their own time. In attempt to balance the need for genetics training with clinicians already busy schedules many suggested using grand rounds, seminars or conferences as a way to incorporate genetics into continuing education.

> What I would love to do is have maybe a quarterly update, you know, maybe…I don't know, maybe rolled into a conference of some kind. You know, one of the conferences that we normally have in Neonatology it might be helpful to have a specifically targeted state of affairs for genetic testing. Here's the update, so now you know. I don't know. (P31, Neonatologist)

Others felt that a return to formal didactics or course-based education is needed.

> "My daughter is taking genetics right now in college and she was starting to go through it and I'm like, oh my god, this is like a whole new world. So I have told her when she is done, we are keeping her textbook and I'm sitting down and reading it." (P1, Cardiologist)

Several felt it would be most valuable for clinicians to have access to genetics education in-real time, as it applied to their patients, an on-the-job version of the case-based learning.

> "Most of us learn best from our patients and so I would think the way to educate people about whole genome sequencing is when they're ordering other stuff that might be supplanted by whole genome sequencing, make that suggestion." (P7, Cardiologist)

Building on this a number of participants looked to the way that genetic results themselves are presented. They commented that results from GS were often so confusing that it was hard to understand what they actually meant in relation to their patient's care, "I don't know how easily we'll be able to pick stuff out that matters." (P9, Intensivist)

A few participants pointed out that simplifying genetic results to make them more interpretable and easier to understand for clinicians could make them more useful.

> "it's got to be simple enough for physicians but there's no person who is a physician who's going to be able to cover, to understand all of that and do their day-to-day job." (P33, Cardiac intensivist)

In addition, clinicians highlighted the nuance in differentiating significant findings from 'clinically significant' findings.

> "say we could move the needle between 40% and 45%, probability and we have a 10% confidence to do that. I don't believe there's any [clinical] utility knowing that information, but if you did know that there was a 20% chance of this fatal disease, then that's a very different equation." (P12, Cardiologist/Geneticist)

DISCUSSION

Our study identifies both gaps and areas for support for clinicians in implementing GS into critical care settings. As a whole participants were concerned about their own inability to accurately use genetic results in clinical decision making, which could have serious impacts on patient care. Consequently, when faced with implementing genomic testing results into high-stakes decisions, interviewed clinicians strongly desired both decision and knowledge guidance from collaboration with genetics specialists, though many felt that such specialists were often not available within the timeframe required by many critical care decisions. Lastly clinicians felt that they needed continuing education for themselves about genomic findings to be able to use GS to improve patient care.

Previous studies have shown disparities in genetic expertise amongst clinicians, especially when it comes to the wide range of results that can come from genomic sequencing (Graf, Char, Hanson-Kahn, & Magnus, 2019). This is especially pertinent for syndromic results that may have implications outside of the ordering clinician's specialty, as well as complicated results such as incidental findings and variants of uncertain significance (VUSs) where there is potential for clinicians to overstate the significance of a finding (Campion, Goldgar, Hopkin, Prows, & Dasgupta, 2019; Graf et al., 2019). The support requested by clinicians in our study was twofold: they desired both general knowledge support in understanding genetic findings for themselves, as well decision support from genetic specialist in contextualizing those results into a patient's care plan and using them in the critical care space.

Many of the clinicians in our cohort expressed being unsure of how to read the genetics results themselves and desired education that would help them be able to actively use genetic information. Clinicians are unlikely to attain the same depth of training as a genetics specialist, such as a genetic counselor, however, they felt it was important to understand the key principles. In this manner, genetic results could be likened to clinicians' understanding of statistics. A cursory understanding of statistics allows physicians to make some clinical decisions based in complex literature, without consultation from a statistician to confirm each studies validity. Improving the clarity of genetics results and training clinicans to use them could allow them to be used in a similar manner.

This highlights the need for the development of systems to communicate the known clinical weight of various findings. The discipline of Evidence-Based Medicine has worked to develop a hierarchical system of classifying evidence for prognostic studies as well as practice recommendations (Burns et al, 2011). In genomics evidence scoring systems, such as ClinGen (https://clinicalgenome.org/), will likely be integral in transitioning genomics into new clinical settings like critical care (Khoury et al., 2018). While bedside clinicians will need to become familiar with these grading scales of findings, having understanding of these scales could streamline their ability to use genetic information and collaborate with genetics professionals on care decisions. Additionally, such rubrics guiding clinicians' interpretation of GS findings might facilitate needed collaborative translational research on the best approaches to implementing genomics to clinical care (Khoury et al., 2018).

Building on this, clinicians in our study stressed a need for further education in genetics and genomics. Further education has tremendous potential to both allow clinicians to make calls on genetic results should a genetics specialist not be available, and work collaboratively with specialists when they are present. This is perhaps the most challenging issue to address, given the significant demands already present on bedside clinicians' time. While formal didactics, like pre-clinical courses in genetics/genomics, would be the most comprehensive they are also the most schedule intrusive (Campion et al., 2019).

Medical education in the last two decades has focused on the value of "case-based learning" (Riddell et al., 2017). Bedside care of an individual patient is what case-based learning aims to duplicate. Prioritization of genetics and genomics education through already existing continuing medical education platforms (such as weekly grand rounds or journal clubs) coordinated with GS implementation might also be beneficial. Additionally, some clinicians desired genetics education to be accessible in real-time as it pertained to patients that they were directly caring for. There could be a role for web-based and self-paced educational modules such as massive open online courses (MOOCS) or other online modules that could provide clinicians with pertinent genetics instruction as it is needed (Campion et al., 2019).

But when it came to the second objective of contextualizing results into a patient's care plan, clinicians saw access to a dedicated critical care genetic counselor as the best solution to addressing their concerns. Stark *et a*l. found similar results in a study of Australian critical care providers who strongly preferred that rapid genomic testing in the acute pediatric setting to be led by clinical genetic services (Stark et al., 2019). Previous studies have highlighted the roles of genetic counselors in critical care settings with a focus on patient

support (Ayres et al., 2019; Clowes Candadai, Sikes, Thies, Freed, & Bennett, 2019). Genetic counselors can allow for long-term follow up and continuity of care surrounding genetic results. They can identify support resources for the family and coordinate cascade testing. They also provide psychosocial support for families in crisis, especially surrounding life-limiting diagnoses and the initiation of palliative care. Genetic counselors also ensure that other implications of genetic testing, such as incidental findings, are considered in high-stress environments (Ayres et al., 2019; Clowes Candadai et al., 2019).

This study highlights yet another important role for counselors in the crucial care setting: working with clinicians to integrate genetic information into critical care decision making. Additionally, increased presence of genetic counselors and a clear rubric for addressing the statistical and evidentiary support behind various GS findings could contribute significantly to on-the-job education of clinicians using and encountering GS. A critical care GC could guide GS testing choices and explain both the findings and their potential clinical implications, not just to the patient or their family, but to the entire care team. Bringing this new perspective to the table will likely enhance discussions around the use of GS in critical care decisions and would provide a point person when clinicians come across unfamiliar information in the genetics space. GCs would also be able to tailor clinician education to be the most fitting for the challenges that emerge in the critical care setting.

Interviewees in this cohort lauded specialized cardiac genetic counselors, when they were available, to provide insight on cardiac-related GS findings. Critical care similarly requires specialty knowledge. GCs serving in such settings would need to have an understanding of the types of decisions being made in critical care, as well as the time pressure that accompany them (Ayres et al., 2019). The desire to have a genetic counseling presence in critical care settings also brings forth the question of if genetic counselors should receive additional formal training to function in critical care settings? There are many unique medical and psychosocial considerations when it comes to counseling in a critical care settings (Ayres et al., 2019; Clowes Candadai et al., 2019; Smith, du Souich, Dragojlovic, Elliott, & Elliott, 2019). In the past genetic counselors have primarily functioned in pediatric, cancer and prenatal settings, but more recently roles in neurology, cardiology, pharmacology and even psychiatry have emerged (Heald et al., 2016). Genetic counseling programs have begun to build specific training in these specialties, as well as topics such as genomic variant interpretation, into their curriculum (Grove et al., 2019). There may be room to incorporate critical care specific training into genetic counseling curriculum or into continuing education programs for genetic counselors.

While most clinicians saw having a specialized critical care genetic counselor as being the gold-standard, they were also aware that there is a scarcity of genetic specialists. It is known that the number of geneticists and genetic counselors is inadequate to interpret results for all patients who undergo GS, even in the outpatient setting (Hoskovec et al., 2018; Ormond et al., 2010). Additionally, one of the key elements of having a genetic counselor in a critical care setting would be having them available to consult within the time-frames of high-pressure critical care decisions and offer continual support to the clinical team. Although it is unclear if a dedicated critical care genetic counselor would be cost-effective, with

significant efforts underway to implement rapid genomic testing to pediatric critical care the need is clear.

Critical care clinicians need support in implementing GS, even if having a fulltime genetic counselor may not be an attainable solution for all hospitals. In lieu of such services there are several additional solutions that could lessen some of the concerns identified by clinicians in our cohort. Hospitals could improve access to genetics consultations through counselors who either work in different departments, or who share responsibilities between multiple hospitals. Tele-genetic counseling is one of the fastest growing areas in genetic counseling and contracting companies could help bring support to clinics on an as needed basis, especially in rural settings, or smaller hospitals (Fang et al., 2016; Stalker et al., 2006).

It is also possible that this is an area where well-designed artificial intelligence (AI) may be able to provide some decision support to clinicians. AI technologies are being developed to assist in interpretation of clinical tests like radiology, and to replace traditional clinical algorithms (Hov et al., 2018; Lynn, 2019; Yoo et al., 2019). AI offers the potential to analyze complicated components of a patient's case which clinicians may not be able to interpret, and it is reasonable to believe that these programs will soon be able to provide guidance on which patients may benefit from genetic testing, and what actions should be taken based on GS results. While AI offers promise to aid in clinical decision making, it is not without limitations and ethical concerns. Algorithms will need substantial training, continual evaluation for accuracy, and may also come with a learning curve before data becomes useful for clinicians.

While these solutions may offer some support and could remove some of the pressures on the already strained genetics workforce, they are unlikely to be able to provide the same benefits for both patients and providers of having a dedicated genetic counselor. The dynamic and evolving nature of implementation of genomic findings to care suggest that, at least for the near term, having an accessible genetic specialist who understands both the current genetic literature as well as the clinical demands of the critical care context is the best approach to support clinical decisions and improve overall clinical knowledge about genomic findings. Hospitals may need a variety of approaches to help clinicians use GS in critical care settings depending on their specific needs and resources. We hope that future research will continue explore ways for hospitals to provide such supports.

LIMITATIONS

The interviewer for all collected interviews (DC) is a practicing clinician who works at the institution. This work relationship may have introduced interviewer bias into the interview dynamic. However, this association may also have provided a dynamic allowing for greater candor by interviewees, such as expressing unguarded anxieties or concerns and ethical challenges that they perceive with GS implementation that they might not have revealed to an interviewer unfamiliar with their work or practice environment. As with all qualitative studies there are limits to generalizability. This study's field site is developing a genomics service, with many clinicians involved in genetic or affiliated research. Clinicians at this

particular field site may have thought about or considered the uses and impacts of GS more than clinicians in other critical care settings.

CONCLUSION

Integrating GS into clinical decision making for critically-ill children with CHD has the potential to improve care and clinical outcomes. For this implementation to be successful, however, clinicians need to have the appropriate training and support to use these results. Our assessment revealed that clinicians do not currently feel comfortable using this information in making high impact decisions. While they felt it was necessary to be able to weigh in on results themselves through continuing genetics education, they saw that having greater presence of genetic counselors in critical care settings was imperative. To best serve in this setting a genetic counselor would need to have an understanding of the complexity and time sensitivity of critical care decisions, as well as context for the psychological needs of the families being evaluated. This presents a new area and important area for genetic counselors to improve care through their unique skill set. Future research is needed to identify best practices for providing this support.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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Table 1:

Demographics of Interviewees

	n	(%)
Gender		
Male	20	57%
Female	15	43%
Relative Seniority (years since completion of training)		
Junior (10 years or less)	12	34%
Mid-Career (10–20 years)	10	29%
Senior (20+ years)	13	37%
Clinician Specialty Type		
Cardiologist	10	29%
• Cardiologist-geneticist (3)		
Nurse Practitioner (NP)	8	23%
Anesthesiologist	6	17%
Intensivist	3	9%
Neonatologist	3	9%
Surgeon	3	9%
• Cardiothoracic (2)		
• Ear nose and throat (ENT) (1)		
Physician Assistant (PA)	2	6%