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Genetics in the NICU: Nurses' Perceived Knowledge and Desired Education

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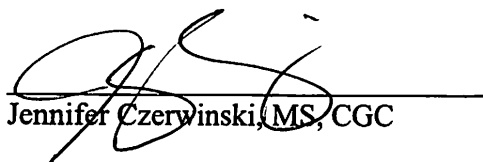
Genetics in the NICU: Nurses' Perceived Knowledge and Desired Education

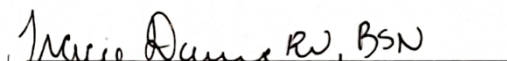
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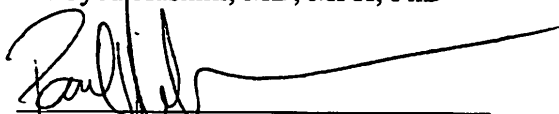


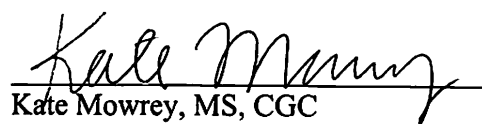
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Genetics in the NICU: Nurses' Perceived Knowledge and Desired Education

A

Thesis

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Genetics in the NICU: Nurses' Perceived Knowledge and Desired Education

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A large proportion of infants admitted to neonatal intensive care units (NICUs) have genetic conditions. NICU nurses play an important role in providing comprehensive care to these patients and their families. Previous research has demonstrated gaps exist in the genetics knowledge of nurses and that they lack comfort applying genetics information to clinical practice, but no research has been done assessing the knowledge of or comfort with genetics of NICU nurses specifically. NICU nurses (n=122) completed an online survey assessing their perceived knowledge of genetics, comfort with clinical scenarios involving genetics, and desired genetics education. Participants reported the highest levels of knowledge of the common trisomies (Down Syndrome, Trisomy 18, Trisomy 13), and genetic testing was a general area of weakness. Over 75% of respondents' overall comfort scores indicated they felt generally uncomfortable with scenarios involving genetics. Perceived knowledge and overall comfort were both impacted by highest degree received, how well prepared a nurse felt by the genetics education they received in their nursing training, and having a close relationship to someone with a genetic condition. Almost all respondents (96%, n=117) desired additional genetics knowledge, with specific interest in genetic conditions encountered in the NICU, genetic testing, and education resources. Gaps exist in the genetics knowledge of neonatal nurses in our cohort, and their overall comfort working with clinical scenarios involving genetics was low. There is a great deal of interest in additional genetics education to support NICU nurses in working with this patient population, and continuing education opportunities should be provided to help them serve this patient population with confidence.

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Introduction

A substantial portion of infants admitted to neonatal intensive care units have genetic conditions, or birth defects suggestive of genetic conditions. This group contributes significantly to mortality in the NICU. In a retrospective review by Malam et al. (2017), 26% of infants admitted to a NICU over a 2-year period were ultimately diagnosed with genetic conditions. A study on the contribution of genetic conditions to NICU mortality found that 28% of the 170 patients who died between 2011 and 2015 had molecularly confirmed genetic diagnoses (Wojcik et al., 2018). It is clear that the burden of genetic disease within the NICU is considerable. Neonatal nurses, who are often the main point of contact for families of patients in the NICU, are well positioned to act as a comforting and knowledgeable liaison between families and physicians (Thorngate & Rios, 2008). These nurses may also be the first to identify dysmorphic features or other signs of genetic conditions that can lead to genetics consultations and diagnoses (Lewis, 2011; Thorngate & Rios, 2008). However, multiple studies have demonstrated that nurses lack knowledge of genetic concepts and have limited confidence in applying genetic principles in clinical care.

In 2005, a consensus panel of key stakeholders from the nursing community worked to establish the *Essential Nursing Competencies and Curricula Guidelines in Genetics and Genomics* (Jenkins & Calzone, 2007). While these guidelines have been an important step towards integrating genetics education into nursing programs, a study evaluating the implementation of the *Essentials* surveyed nurses on the curriculum content at their schools and found that most programs at both the baccalaureate and graduate level do not fully meet these competencies, 78.7% and 85.1% respectively (Thompson & Brooks, 2011). Studies of the perceived knowledge and attitude towards genetics in both undergraduate and graduate nursing students revealed that these students lack knowledge of medical genetics and lack the comfort to integrate this information into practice (Maradiegue et al., 2005; Dodson & Lewallen, 2011). A

study by Calzone et al 2013 showed that while the majority of nurses surveyed consider genetics and genomics important to nursing practice, 63% reported that their overall knowledge of genetics and genomics was poor or fair. These studies have all demonstrated that gaps exist in genetics knowledge within the nursing community.

Previous studies have primarily focused on nurses' knowledge and comfort with the genetics of common diseases, like cancer, heart disease, and diabetes (Calzone et al., 2012; Calzone et al., 2013). Fewer studies have considered nurses' knowledge of and confidence in working with genetic conditions more often seen in the NICU setting. There have, however, been studies on nursing students' perceived knowledge of these types of conditions and their level of integration into nursing curricula. Dodson & Lewallen (2011) found that most nursing students reported having no or minimal knowledge of conditions like osteogenesis imperfecta, phenylketonuria, and trisomies 13, 18, and 21. In one study examining the integration of genetic concepts into nurse practitioner curricula, it was found that over half of faculty were not comfortable teaching about conditions like myotonic dystrophy, Klinefelter syndrome, cystic fibrosis, and the common trisomies (Edwards et al., 2006). To our knowledge, a study to assess the medical genetics knowledge of NICU nurses specifically has not yet been performed. Because neonatal nurses play such a critical role in providing comprehensive care to NICU patients and may be the first members of the health care team parents can go to with questions about their child's condition and care, it is important to learn more about NICU nurses' knowledge of and comfort with genetics concepts.

The purpose of this study is to describe the perceived medical genetics knowledge of nurses working in NICUs and their level of comfort with working through clinical scenarios involving genetics concepts. This study also assessed what further genetics education is desired so that in the future educational resources can be developed to support NICU nurses in their work caring for patients with genetic conditions.

Methods

Study design

This study was performed using a cross-sectional approach to survey nurses working in level 3 and 4 NICUs. The electronically delivered, anonymous survey was designed using the Qualtrics online software. The survey was developed by the research team. Approval was obtained from the Institutional Review Board of University of Texas Health Science Center at Houston (HSC-MS-20-0635), the Memorial Hermann Healthcare System, and the National Association of Neonatal Nurses (NANN) Research Committee. The survey was distributed via email to nurses at the Children's Memorial Hermann Hospital (CMHH) NICU, and was also available through the NANN newsletter and the MyNANN Community message boards. Data was collected between September 2020 and January 2021. Prior to completing the anonymous survey, all participants provided consent electronically. Upon completion of the survey, participants were given the option to provide contact information for a chance to win one of four \$25 gift cards.

Study Sample

The inclusion criteria for participating in this study was working as a registered nurse in level 3 or 4 NICUs. Participants were recruited from the Children's Memorial Hermann Hospital (CMHH) NICU, a level 4 NICU in Houston, TX, and from the members of the National Association of Neonatal Nurses (NANN). The CMHH NICU employs approximately 180 NICU nurses. NANN currently has approximately 8,000 members from across the United States, working in all neonatal nursing settings. The recruitment materials shared with NANN and the electronic consent form completed by all participants prior to beginning the study survey included the inclusion criterion of currently working in a level 3 or 4 NICU.

Survey Development

The survey used in this study collected basic demographic and education information, assessed nurses' perceived knowledge of genetics, their comfort with clinical scenarios involving genetics, and what genetics education would be helpful to them in their work in the NICU. The survey was developed by the authors and included questions on perceived knowledge adapted from the survey tool used by Maradiegue et al., 2005, and one question taken from the instrument in the "Survey of Nursing Integration of Genomics Into Nursing Practice" (Calzone et al., 2012). The first section of the survey collected demographic information and information related to the nurse's training and practice history. The second section of the survey was an assessment of perceived genetics knowledge. In this section, perceived knowledge was evaluated using a four-point Likert scale ranging from "No knowledge of this topic" to "High level of knowledge of this topic." There were 26 genetics topics assessed. These included basic genetics concepts, genetic conditions frequently encountered in the NICU population, and forms of genetic testing. The next section of the survey was an assessment of respondent's level of comfort working through different clinical scenarios related to genetics. There were eight scenarios, and respondents indicated their level of comfort using a sliding bar on a 0-100 scale from "Extremely Uncomfortable" to "Extremely Comfortable," with any rating over 50 indicating some degree of comfort. These scenarios, listed in Table 1, were developed by the authors with some of the specific knowledge areas and clinical performance indicators included in the document "Establishing the Outcome Indicators for the Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics" in mind (Calzone et al., 2011). Finally, the survey assessed desired genetics education and how valuable the respondents perceived genetics knowledge is to their work in the NICU. There were six topics, and respondents selected how helpful they feel an understanding of that topic is

to their work in the NICU from a five-point Likert scale ranging from “Not helpful” to “Very helpful.” Respondents were also asked if they desire further genetics education. Those who selected yes were then asked what three genetics topics they want to better understand (free response), and how they would like that education delivered.

Table 1. List of clinical scenarios involving genetics.

Clinical Scenarios Involving Genetics	
Scenario 1	The parents of one of your patients who was just diagnosed with cystic fibrosis ask you to explain the recurrence risk of this autosomal recessive condition. How comfortable do you feel explaining this inheritance pattern to the family?
Scenario 2	You have a patient who is newly diagnosed with Down syndrome. You see the family struggling to find information and resources. How comfortable do you feel identifying information and resources about their baby’s genetic condition?
Scenario 3	One of your patient’s features are suspicious for a genetic condition. How comfortable do you feel taking a pedigree and gathering a thorough family history?
Scenario 4	The chromosome microarray (CMA) results for one of your patients have just returned. How comfortable do you feel understanding the test report?
Scenario 5	You have a new patient with dysmorphic facial features. How comfortable do you feel conducting a physical assessment that incorporates knowledge about genetic, environmental, and genomic influences and risk factors?
Scenario 6	The parents of one of your patients who has been diagnosed with Down syndrome believe that they can “pray away” the baby’s condition. How comfortable do you feel discussing the permanency of this genetic condition?
Scenario 7	You have a patient with many features suggestive of a genetic condition who had a normal CMA. The parents therefore believe that this is not genetic and don’t understand why further testing is being recommended. How comfortable do you feel helping clarify why additional tests are being recommended?
Scenario 8	Your patient, who was diagnosed with CHARGE Syndrome, is preparing for discharge. The family has many questions related to this genetic condition as they get ready to go home. How comfortable do you feel providing education to this family?

Statistical analysis

All data were extracted from Qualtrics and data analysis was performed using Stata (v. 13.1, College Station, TX, USA). Descriptive statistics were used to report all variables of interest. Mean perceived knowledge and mean comfort scores were calculated for each participant by averaging their responses to the 26 genetics topics and the eight clinical scenarios to determine an overall perceived knowledge and comfort score for each participant. Using these mean knowledge and comfort scores, distributions of continuous variables were compared using either a Kruskal-Wallis test with post-hoc Dunn test (for comparisons between more than two groups) or a Mann-Whitney test (for comparison between two groups).

Free response questions on desired genetics education were analyzed using latent content analysis. Latent content analysis is a method designed to identify and interpret meaning in free response questions by identifying individual themes in order to describe or explain a theoretical framework (Bengtsson, 2016; Down-Wamboldt 1992). The primary and last author independently categorized each free response into one or more identified themes, with the thematic coding for each response subsequently compared and agreed upon.

Results

Demographics

During the study period, 129 neonatal nurses completed the online survey. Of these, seven were excluded due to completing less than 70% of the survey. The remaining 122 participants were included in final data analysis. The average age of respondents was 40.3 years (sd 13.1). The age group most represented in this cohort was under the age of 35 (44%, n=54). Participants' demographics, education, and experience are summarized in Table 2.

Characteristics	n (%) n=122
Age	
23-34	54 (44%)
35-49	29 (24%)
50 +	39 (32%)
Highest Degree	
BSN	92 (75%)
MSN	17 (14%)
DNP	1 (1%)
PhD	1 (1%)
Other	11 (9%)
Graduation Year (Highest Degree)	
<2011	52 (43%)
2011-2015	29 (24%)
2016+	41 (33%)
Years in nursing	
<5 years	36 (30%)
6-10 years	20 (16%)
11-15 years	16 (13%)
16-20 years	9 (7%)
21-30 years	19 (16%)
>30 years	22 (18%)
Years in NICU	
<5 years	39 (32%)
6-10 years	19 (16%)
11-15 years	21 (17%)
16-20 years	14 (11%)
21-30 years	13 (11%)
>30 years	16 (13%)
NANN Member	
Yes	58 (48%)
No	64 (52%)

Table 2. Characteristics of participants.

Education, Experience, and Exposure to Genetics

The majority of study participants' highest degree was a BSN (75%, n=92), followed by 14% who had an MSN (n=17). One participant each had a DNP and PhD, and 9% of respondents reported having a different highest degree (n=11). Most participants reported that their nursing training did incorporate genetics education (68%, n=83). Most often, participants reported that genetics topics were discussed in lectures on other topics (57.8%, n=48), but about one-third reported receiving at least one dedicated lecture on genetics (31%, n=26) and 8% (n=7) took a dedicated course on genetics. When asked how well prepared they felt by the genetics education the received, 29% (n=24) did not feel well prepared, 70% (n=58) felt somewhat prepared, and a single respondent (1%) felt well prepared (1%). Very few participants reported an awareness of the *Essential Competencies and Curricula Guidelines for Nurses in Genetics and Genomics* (5%, n=6).

Approximately one-third of respondents have worked in nursing or the NICU for less than five years (30%, n= 36 and 32%, n=39 respectively). Most respondents did not report having worked in another nursing specialty prior to working in the NICU (64%, n=78). Just over three-quarters of participants work in a NICU alongside a genetics consult team (77%, n=94).

All but one respondent reported that they had participated in the care of a patient with a genetic condition (99%, n=121). About one-tenth of participants reported personally having a genetic condition (9%, n=11), and about one-third reported having a family member or close friend with a genetic condition (29%, n=35).

Perceived Knowledge and Comfort with Clinical Scenarios Involving Genetics

To evaluate perceived knowledge, participants in this survey were asked to report their current knowledge of 26 topics related to genetics. The results of this section are illustrated in Figure 1. The topics participants reported having the highest level of knowledge in were Down syndrome, trisomy 18, and trisomy 13 with 96% (n=117), 82% (n=100), and 75% (n=92%) reporting some or high levels of knowledge of these topics respectively. Nurses reported lower levels of perceived knowledge in topic areas related to genetic testing. About half of participants reported some or high levels of perceived knowledge of chromosomal microarrays (CMA) and karyotypes (56%, n= 67 and 49%, n=60 respectively), but a majority reported no or some perceived knowledge of single gene testing, whole genome sequencing (WGS), and whole exome sequencing (WES) (73%, n=8; 79%, n=96; and 70%, n=85 respectively).

Perceived Genetics Knowledge

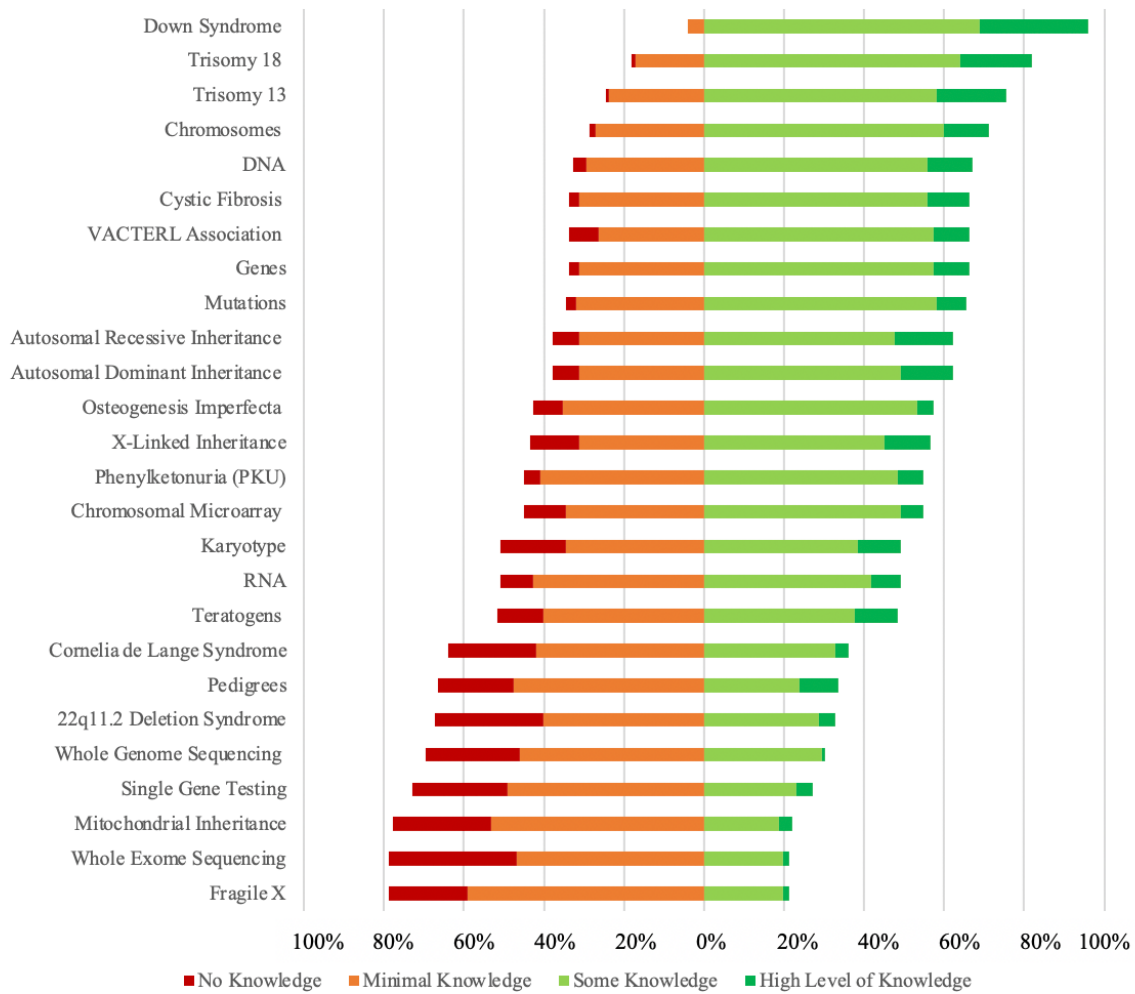


Figure 1. Participants' perceived knowledge of 26 genetics topics.

To assess participants' comfort working with genetics information, they were asked to indicate how comfortable they would be working in eight different clinical scenarios involving genetics. Figure 2 illustrates the results from this section. The scenario with the highest mean comfort was one involving explaining the permanency of a genetic condition to a family that hoped to "pray away" the diagnosis (mean=68.1, sd=29.5). The only other scenario with a mean rating over 50 was one assessing comfort identifying resources for a family whose baby was diagnosed with Down syndrome (mean=61.6, sd=26.7). The scenarios with the lowest mean ratings were those related to taking a pedigree and family history, understanding a patient's

CMA report, and providing education to a family related to their child's diagnosis prior to discharge (mean=23.0, sd=23.5; mean=30.2, sd=26.2; and mean=29.4, sd=27.1, respectively).

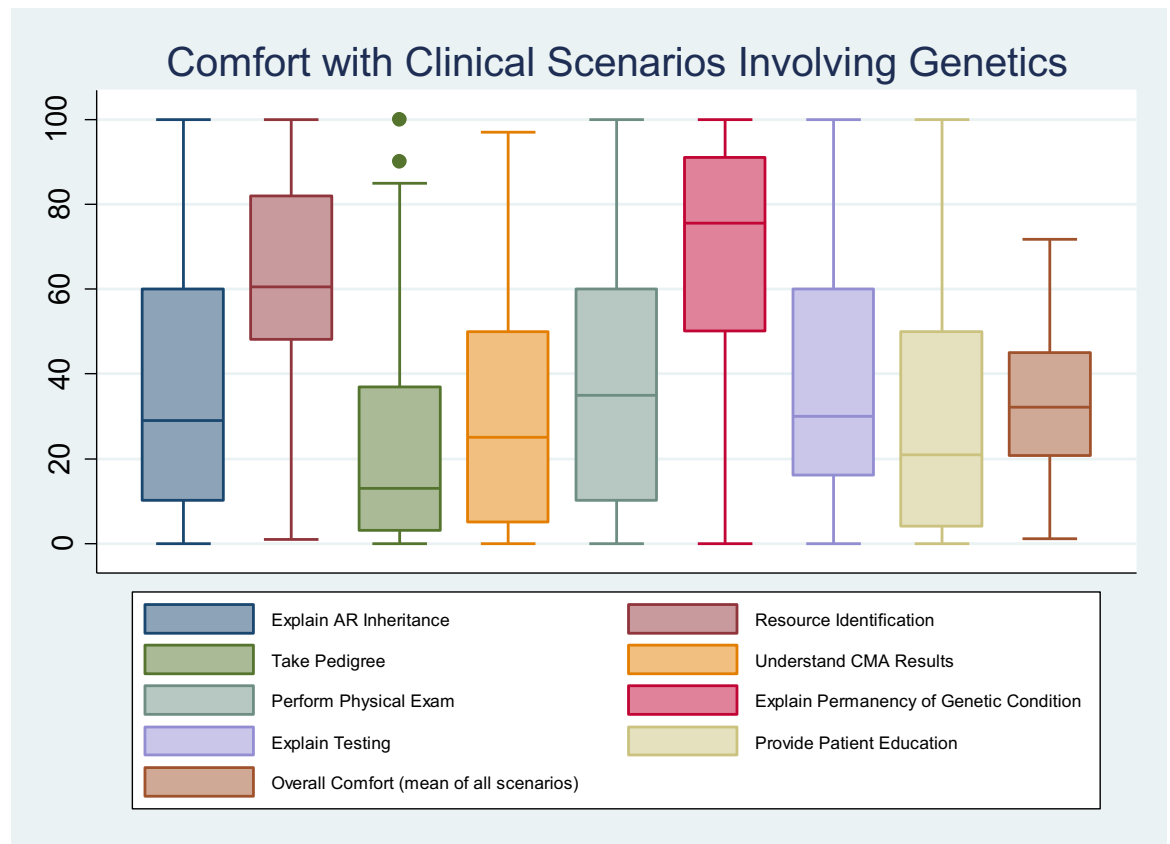


Figure 2. Participants' comfort with clinical scenarios involving genetics.

Similar factors impacted both participants' perceived knowledge and level of comfort with the clinical scenarios (Tables 3 and 4). There was no statistically significant difference in the mean knowledge between the three age groups ($p=0.774$). However, a statistically significant difference was observed for highest degree attained in perceived knowledge ($p=0.030$). Respondents with a BSN degree had a lower median perceived knowledge score (median: 2.46; IQR: 2.17 - 2.73) compared to those with an MSN (median 2.77; IQR: 2.35 - 3.00). The single respondents who reported having a DNP or PhD also had a higher score (3.88 and 3.35, respectively) than the mean scores for MSN. The perceived knowledge score varied significantly by how prepared a nurse felt by the genetics education they received ($p=0.001$). The median perceived knowledge score was 2.23 (IQR: 1.98-2.46) among respondents who

indicated they did not feel prepared by their genetics training. The median perceived knowledge score was 2.60 (IQR: 2.35-2.85) among respondents who indicated they felt somewhat prepared by their genetics training. There was a single respondent who indicated that they felt well prepared by their genetics training, and their mean perceived knowledge score was 3.88. Finally, a statistically significant difference in perceived knowledge scores was found between participants who did and did not have a family member or close friend with a genetic condition ($p=0.000$). Respondents who are closely related to someone with a genetic condition had a median knowledge score of 2.73 (IQR: 2.46-3) compared to a median score of 2.35 (IQR: 2.15-2.65) for those who are not.

A statistically significant difference in comfort was found between the three age groups ($p=0.028$). Participants from the 35-49 age group had a median comfort score of 46.31 (24.45-51.77), higher than the median scores for the groups <35 and 50+ (27.86, IQR: 13.83-41.71 and 29.56, IQR: 20.7-44.58 respectively). The comfort score was significantly different when stratified by highest degree ($p=0.030$). The median comfort score was 44.89 (IQR: 29.56-55.64) among respondents with an MSN and was 28.81 (IQR: 18.15-42.79) among those with a BSN. A statistically significant difference was also observed based on how well prepared a respondent felt by the genetics education they received during their nursing training ($p=0.003$). The median comfort score was 24.00 (IQR: 12.22-33.83) among respondents who indicated they did not feel prepared by their genetics training. The median comfort score was 36.34 (IQR: 24.53-47.53) among respondents who indicated they felt somewhat prepared by their genetics training. There was a single respondent who indicated that they felt well prepared by their genetics training, and their mean comfort score was 69.19. As with knowledge, a statistically significant difference was found in comfort between respondents with and without a close relationship to someone with a genetic condition ($p=0.012$). The median comfort score among respondents who have a

close friend or family member with a genetic condition was 41.8 (IQR: 23.55-49.83) compared to 27.66 (IQR: 17.99-42.56) for those who do not.

Table 3. Relationships between variables and overall knowledge.

Knowledge					
	Count	Median (IQR)	p-value	Crude OR	Adjusted OR
Age Group					
<35	54	2.52 (2.23 - 2.73)	0.7737	Ref	
35-49	29	2.46 (2.27 - 2.85)		0.06 (-0.17 , 0.29)	-0.01 (-0.58 , 0.57)
>50	39	2.46 (2.19 - 2.77)		-0.06 (-0.27 , 0.15)	0.1 (-0.58 , 0.77)
Degree					
BSN	92	2.46 (2.17 - 2.73)	0.0289	Ref	
MSN	17	2.77 (2.35 - 3)		0.34 (0.09 , 0.59)	0.28 (-0.05 , 0.61)
DNP	1	3.88 (3.88 - 3.88)		1.44 (0.49 , 2.4)	2.61 (1.29 , 3.93)
PhD	1	3.35 (3.35 - 3.35)		0.91 (-0.05 , 1.86)	0.54 (-0.43 , 1.51)
Other	11	2.38 (2.23 - 2.54)			-0.12 (-0.5 , 0.25)
Grad Year					
<2011	52	2.46 (2.21 - 2.79)	0.9685	Ref	
2011-2015	29	2.54 (2.27 - 2.65)		0.09 (-0.14 , 0.33)	0.42 (-0.06 , 0.9)
2016+	41	2.46 (2.23 - 2.81)		0.09 (-0.12 , 0.3)	0.16 (-0.31 , 0.64)
Genetics Education					
No	39	2.46 (2.15 - 2.77)	0.6386	0.08 (-0.11 , 0.28)	
Yes	83	2.46 (2.23 - 2.81)		2.45 (2.29 , 2.61)	
Preparedness					
Not prepared	24	2.23 (1.98 - 2.46)	0.0007	Ref	
Somewhat prepare	58	2.6 (2.35 - 2.85)		0.36 (0.15 , 0.58)	0.3 (0.06 , 0.55)
Well prepared	1	3.88 (3.88 - 3.88)		1.63 (0.72 , 2.53)	0 (omitted)
Years in Nursing					
<5	36	2.5 (2.19 - 2.83)	0.9759	Ref	
6-10	20	2.44 (2.25 - 2.69)		-0.05 (-0.33 , 0.24)	0.51 (-0.5 , 1.51)
11-15	16	2.5 (2.17 - 2.75)		-0.05 (-0.35 , 0.26)	0.6 (-0.72 , 1.92)
16-20	9	2.35 (2.23 - 2.46)		-0.09 (-0.47 , 0.29)	0.2 (-1.17 , 1.56)
21-30	19	2.38 (2.23 - 2.88)		-0.12 (-0.41 , 0.17)	0.1 (-1.28 , 1.48)
30+	22	2.54 (2.27 - 2.77)		0.01 (-0.27 , 0.29)	-0.29 (-1.8 , 1.22)
Years in NICU					
<5	39	2.46 (2.15 - 2.85)	0.7042	Ref	
6-10	19	2.5 (2.23 - 2.65)		-0.14 (-0.42 , 0.14)	-0.75 (-1.71 , 0.21)
11-15	21	2.46 (2.19 - 2.85)		0.04 (-0.23 , 0.31)	-0.44 (-1.83 , 0.95)
16-20	14	2.44 (2.27 - 2.65)		0.02 (-0.3 , 0.33)	0.03 (-1.37 , 1.43)
21-30	13	2.35 (1.92 - 2.62)		-0.29 (-0.61 , 0.03)	0.07 (-1.38 , 1.52)
30+	16	2.63 (2.31 - 2.79)		0.06 (-0.24 , 0.36)	0.6 (-0.96 , 2.15)
Other Specialties					
No	78	2.46 (2.31 - 2.77)	0.3948		
Yes	44	2.4 (2.15 - 2.75)		-0.03 (-0.22 , 0.16)	-0.13 (-0.42 , 0.15)
Patient with GC?					
No	1	1.58 (1.58 - 1.58)	0.2033		
Yes	121	2.46 (2.23 - 2.77)		0.94 (-0.06 , 1.94)	1.07 (0.14 , 2)
Personal GC?					
No	111	2.46 (2.23 - 2.77)	0.1007		
Yes	11	2.65 (2.58 - 2.77)		0.23 (-0.08 , 0.55)	0.1 (-0.24 , 0.44)
Fam/Friend GC?					
No	87	2.35 (2.15 - 2.65)	0.0002		
Yes	35	2.73 (2.46 - 3)		0.31 (0.11 , 0.5)	0 (-0.24 , 0.23)
Genetics Consult team					
No	28	2.37 (2.25 - 2.65)	0.7376		
Yes	94	2.46 (2.23 - 2.77)		0.01 (-0.21 , 0.23)	0.12 (-0.15 , 0.39)
Aware of Essentials					
No	116	2.46 (2.23 - 2.77)	0.4069		
Yes	6	2.71 (2.15 - 3.27)		0.32 (-0.1 , 0.74)	-0.14 (-0.72 , 0.44)
NANN Member					
No	64	2.44 (2.15 - 2.71)	0.1176		
Yes	58	2.54 (2.27 - 2.81)		0.18 (0 , 0.36)	0.11 (-0.13 , 0.35)

Table 4. Relationships between variables and overall comfort.

		Comfort				
		Count	Median (IQR)4	p-value5	Crude OR6	Adjusted OR7
Age Group						
	<35	54	27.86 (13.83 - 41.71)	0.0283	Ref	
	35-49	29	46.31 (24.45 - 51.77)		10.16 (2.41 , 17.91)	20.82 (0.81 , 40.82)
	>50	39	29.56 (20.7 - 44.58)		2.64 (-4.43 , 9.72)	20.56 (-3.06 , 44.17)
Degree						
	BSN	92	28.81 (18.15 - 42.79)	0.0292	Ref	
	MSN	17	44.89 (29.56 - 55.64)		13.91 (5.23 , 22.6)	12.1 (0.58 , 23.61)
	DNP	1	69.19 (69.19 - 69.19)		38.51 (5.44 , 71.57)	68.12 (22.04 , 114.2)
	PhD	1	34.93 (34.93 - 34.93)		4.25 (-28.82 , 37.32)	-13.89 (-47.76 , 19.99)
	Other	11	24.45 (20.7 - 50.71)		0.64 (-9.86 , 11.13)	-2.83 (-15.91 , 10.25)
Grad Year						
	<2011	52	28.2 (20.57 - 46.12)	0.7501	Ref	
	2011-2015	29	33.37 (26.03 - 41.72)		0.23 (-7.77 , 8.22)	16.19 (-0.7 , 33.08)
	2016+	41	34.07 (21.52 - 48.85)		3.31 (-3.9 , 10.51)	10.36 (-6.32 , 27.05)
Genetics Education						
	No	39	34.07 (13.02 - 43.32)	0.7171	1.16 (-5.53 , 7.85)	
	Yes	83	31.23 (21.52 - 45.93)		32.24 (26.72 , 37.76)	
Preparedness						
	Not prepared	24	24 (12.22 - 33.83)	0.0026	Ref	
	Somewhat prepare	58	36.34 (24.54 - 47.53)		11.7 (4.28 , 19.13)	9.85 (1.41 , 18.29)
	Well prepared	1	69.19 (69.19 - 69.19)		44.5 (13.28 , 75.72)	0 (omitted)
Years in Nursing						
	<5	36	33.31 (20.03 - 44.02)	0.7194	Ref	
	6-10	20	27.86 (12.58 - 41.32)		-4.86 (-14.53 , 4.81)	9.16 (-25.99 , 44.32)
	11-15	16	37.02 (23.92 - 46.08)		2.96 (-7.46 , 13.38)	14.14 (-32.07 , 60.35)
	16-20	9	46.31 (20.71 - 51.73)		4.89 (-8.03 , 17.81)	-0.47 (-48.27 , 47.34)
	21-30	19	35.86 (20.43 - 46.9)		-0.76 (-10.59 , 9.08)	-20.3 (-68.63 , 28.03)
	30+	22	28.2 (23.55 - 44.78)		0.42 (-8.96 , 9.81)	-26.99 (-79.93 , 25.96)
Years in NICU						
	<5	39	33.25 (17.96 - 44.89)	0.3067	Ref	
	6-10	19	28.05 (12.44 - 37.47)		-5.01 (-14.55 , 4.53)	-16.33 (-50.08 , 17.43)
	11-15	21	34.93 (23.38 - 48.85)		5.04 (-4.19 , 14.27)	-21.79 (-70.44 , 26.86)
	16-20	14	43.46 (20.71 - 51.77)		7.53 (-3.1 , 18.16)	-0.98 (-49.96 , 47.99)
	21-30	13	23.55 (20.43 - 43.03)		-3.9 (-14.83 , 7.02)	10.68 (-40.14 , 61.49)
	30+	16	28.2 (24.05 - 45.35)		1.99 (-8.14 , 12.12)	17.67 (-36.77 , 72.1)
Other Specialties						
	No	78	28.49 (20.43 - 43.32)	0.3704		
	Yes	44	34.96 (21.36 - 47.51)		3.13 (-3.34 , 9.61)	6.42 (-3.54 , 16.38)
Patient with GC?						
	No	1	24.46 (24.46 - 24.46)	0.5798		
	Yes	121	33.25 (20.7 - 45.08)		8.64 (-25.94 , 43.23)	13.05 (-19.5 , 45.61)
Personal GC?						
	No	111	28.93 (20.42 - 44.89)	0.0611		
	Yes	11	39.86 (30.57 - 53)		10.01 (-0.74 , 20.76)	5.18 (-6.67 , 17.04)
Fam/Friend GC?						
	No	87	27.66 (17.99 - 42.56)	0.0121		
	Yes	35	41.48 (23.55 - 49.83)		9.11 (2.4 , 15.81)	-0.96 (-9.24 , 7.33)
Genetics Consult team						
	No	28	28.93 (20.42 - 44.89)	0.7864		
	Yes	94	39.86 (30.57 - 53)		0.9 (-6.52 , 8.32)	6.49 (-2.98 , 15.97)
Aware of Essentials						
	No	116	30.9 (20.7 - 44.83)	0.6443		
	Yes	6	42.37 (10.71 - 48.85)		4.19 (-10.23 , 18.61)	8.39 (-11.99 , 28.77)
NANN Member						
	No	64	27.6 (20.7 - 42.68)	0.1512		
	Yes	58	36.6 (20.43 - 47.53)		4.93 (-1.26 , 11.12)	5.44 (-2.96 , 13.83)

A moderate to good positive correlation was found between participants' mean perceived knowledge and mean comfort scores ($p=0.000$, Pearson's correlation coefficient=0.594), as illustrated in Figure 3.

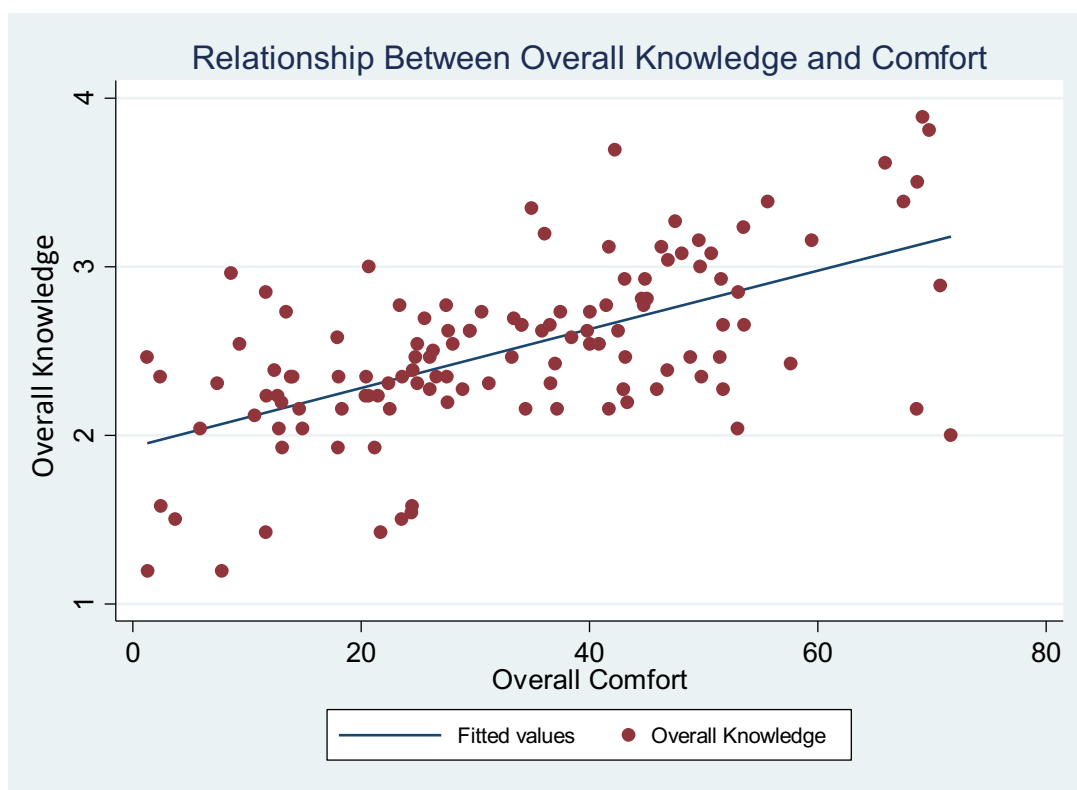


Figure 3. Relationship between overall knowledge and comfort.

Desired Genetics Education

In the assessment of desired genetics education, 96% (n=117) of respondents indicated that they do desire further genetics education. When asked how they would like to receive this education, the most popular response was through lectures (86%, n=100). Participants were also asked to list the top three genetics topics they would like to better understand. Ninety participants completed this question. When reviewing these responses, eight main themes emerged, including genetic conditions encountered in the NICU (n=51), genetic testing (n=45), educational resources (n=32), assessment for genetic conditions (n=18), resources for parent/family support (n=11), basic genetics concepts (n=8), family history/pedigree (n=8), and availability of treatment (n=4). Themes from these responses are summarized in Table 4. When asked how helpful an understanding of six different genetics topics is to work in the NICU, all

six topics were rated at least moderately helpful by a majority of participants. These results are summarized in Figure 4.

Table 5. Excerpts and thematic analysis of free responses.

Themes	Participant Responses
Genetic conditions encountered in the NICU (n=51)	"I would love learning about various genetic disorders, prognosis, and clinical features of each. "
Genetic testing (n=45)	"I would like to know more about the tests we run and what we are looking for."
Educational resources (n=32)	"Resources on where to learn about more rare genetic conditions both for medical professionals and families."
Assessment for genetic conditions (n=18)	"What physical features should I be worried about and present to the physician? "
Resources for parent/family support (n=11)	"Compassionate interactions with families as they are learning of their baby's disorder(s) and how to then care for their baby while they are grieving the loss of the "normal" baby they are not taking home."
Basic genetics concepts (n=8)	"Understanding DNA & deletions/additional chromosomes." "Mosaicism." "Phenotype vs. genotype."
Family history/pedigree (n=8)	"Information on pedigrees, inheritance patterns of certain conditions."
Availability of treatment (n=4)	"Gene therapies that are currently available & how they will change our ideas regarding prognosis (for example Spinal Muscle Atrophy)."

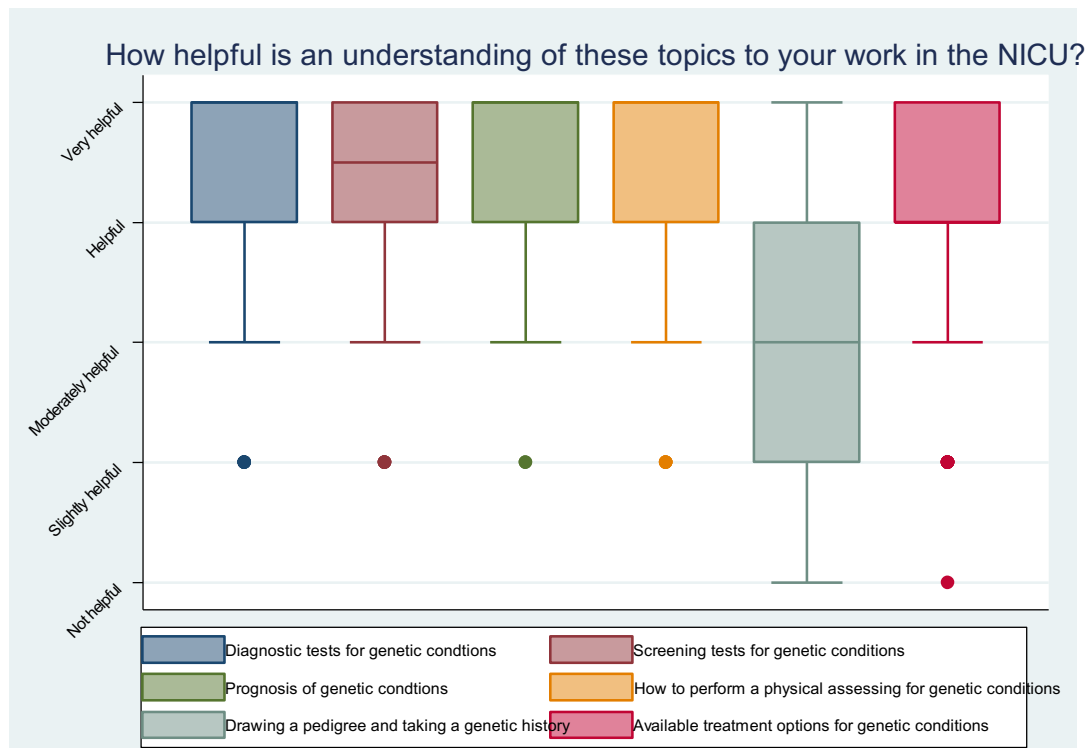


Figure 4. Helpfulness of various genetics topics to work in NICU.

Discussion

This study explored neonatal nurses' perceived genetics knowledge, level of comfort working through clinical scenarios involving genetics, and desired genetics education. Our results build on existing evidence of gaps in the genetics knowledge of nurses and low levels of comfort applying genetics information to nursing practice. These findings contribute a clearer understanding of what factors may influence knowledge and comfort, and provide greater detail on the genetics education neonatal nurses desire.

Participants reported the greatest levels of knowledge of the common trisomies (Down syndrome, trisomy 18, and trisomy 13), three genetic conditions frequently encountered in the NICU setting. Most respondents also reported some or high levels of knowledge of some basic genetics concepts, like DNA, mutations, and inheritance patterns, which is consistent with findings in previous studies (Maradigue et al., 2005; Dodson & Lewallen 2011). A general area of weakness in perceived knowledge our study identified was genetic testing. Although roughly half of participants reported some or high levels of knowledge of CMAs and karyotypes, a majority reported minimal or no knowledge of WES, WGS, and single gene testing. While these tests are newer than CMA and karyotype, which could contribute to the low levels of perceived knowledge, it is clear from free response answers on desired genetics education that participants are very interested in learning more about various forms of genetic testing. Additionally, even though just over half of participants reported some or high levels of knowledge of CMA, the scenarios involving understanding a CMA report and explaining the need for further testing after a normal CMA had low median comfort scores (25 and 30, respectively), indicating that more than half of participants felt uneasy applying their knowledge of CMA in clinical practice.

Overall, our results also demonstrated that most neonatal nurses felt uncomfortable applying genetics knowledge when working through various clinical scenarios involving genetics, with over 75% of participants scoring below a 50 for their overall comfort scores and

only two of eight scenarios with median ratings over 50. This is consistent with the findings of Maradigue et al's study that found most graduate-level advanced practice nursing students felt uncomfortable applying medical genetics concepts to clinical practice (2005). Both of the most highly rated scenarios were related to working with a patient diagnosed with Down syndrome, which was the genetics concept respondents reported the highest level of perceived knowledge of. This is in line with our finding of a moderate to good positive correlation between knowledge and comfort, which highlights the need to offer further genetics education to neonatal nurses. By providing opportunities to improve neonatal nurses' knowledge of genetics, their overall comfort working with this information will also increase, which would allow them to serve this patient population with greater confidence and ease.

Interestingly, there was no significant relationship between the length of time working as a nurse or in the NICU and a respondent's reported perceived genetics knowledge or comfort with genetics. Additionally, our results found no significant difference between perceived knowledge or comfort regardless of whether or not respondents worked in a NICU with a genetics consult team. These results would indicate that exposure to patients with genetic conditions in clinical practice alone, which all but one of our participants report experiencing, and working alongside specialists with genetics expertise is not what increases nurses' perceived knowledge of or comfort with genetics information. Instead, our results suggest that the genetics education nurses receive in their nursing training has a more significant impact on their knowledge and comfort than nursing experience, with nurses with advanced degrees and who felt somewhat or well prepared by their genetics education reporting higher levels of perceived knowledge and comfort. These results demonstrate the importance of including genetics education in nursing curriculum, as well as the value of offering continuing education on genetics to nurses.

The vast majority of participants in our study desired further genetics education (96%, n=117). The topics respondents were most interested in learning more about were genetic conditions encountered in the NICU, genetic testing, and educational resources. Our results demonstrated a high level of interest in having further education delivered via recorded lectures and continuing education unit (CEU) courses that could be completed on a nurse's own time. In future studies, a genetics focused educational intervention for NICU nurses could be developed, with participants' knowledge and comfort measured before and after completion.

One area our study did not explore is what neonatal nurses perceive their role to be in participating in the care of a patient undergoing a genetics workup. While neonatal nurses are often the first touch point for a family and act as a liaison to the physicians and other specialists, their role in patient care certainly is different from that of a geneticist or genetic counselor. While our study has provided new insight into NICU nurses' comfort with genetics and their perceived and desired genetics knowledge, future studies could explore what these nurses believe their role is or should be in caring for this patient population.

Limitations

There are several limitations to our study design and the results. Due to the fact that there is overlap between the populations we sampled (NICU nurses at CHMM and members of NANN), we cannot rule out the of duplicated responses. Additionally, there is likely a selection bias, with neonatal nurses who have an interest in genetics possibly more likely to participate than others. Given this as well as the small sample size, we recognize that our sample population may not be representative of the entire population of neonatal nurses. Our data also included an outlier in our single participant with a DNP, who was also the only individual who indicated they felt well prepared by the genetics education they received during their nursing training. This individual also scored the highest of all participants in their overall perceived

knowledge and comfort scores (3.88 and 69.19 respectively). Finally, the survey tool used in this study is not validated, and the knowledge scores are based on perception rather than assessment.

Future Directions

As also recommended above, future studies could evaluate the impact of a genetics focused educational intervention on the perceived knowledge of and comfort with genetics of neonatal nurses, as well as exploring nurses' understanding of their role in caring for patients undergoing a genetics evaluation. In addition, future research is needed to objectively assess neonatal nurses' genetics knowledge. Further studies should also consider patient satisfaction by exploring the perceptions of parents whose children are included in this patient population.

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Vita

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