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How Neonatologists Use Genetic Testing: Findings from a national survey

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INTRODUCTION

Genetic testing is increasingly common in the neonatal intensive care unit (NICU) given the high prevalence of genetic conditions and risks,[1] but how neonatologists use testing remains unclear. Critics have warned that the frequency and complexity of genetic tests may outpace the necessary supports to render results useful.[2] We therefore conducted a national survey of neonatologists regarding practices for genetic testing and counseling.

METHODS

In the fall of 2020, we emailed a 16-item questionnaire on how neonatologists use genetic tests to approximately 3 600 neonatologists through the listservs of the American Academy of Pediatrics Section on Neonatal and Perinatal Medicine (SONPM) and the Children's Hospitals Neonatal Consortium (CHNC). Data were collected and managed using REDCap tools hosted at the Children's Hospital of Philadelphia (CHOP).

We developed the questionnaire through literature review and consultation with geneticists and neonatologists at level III and IV NICUs and pilot-tested the questionnaire with ten experienced neonatologists. The questionnaire contained multiple-choice questions on use and counseling surrounding genetic testing and a single free-response question inviting additional thoughts on how neonatologists use genetic results in clinical practice. We performed descriptive analyses of multiple-choice responses using Stata version 16.1 (StataCorp, College Station, TX). Free responses were coded by two investigators (K.P.C.,

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Author Information

K.P.C. conceptualized and designed the study, created the study instrument, collected and analyzed the data, drafted the initial manuscript, and reviewed and revised the manuscript. C.F. assisted in designing the study, creating the study instrument, analyzing the data, and reviewed and revised the manuscript. S.J, D.M, K.T.W, and C.S. assisted in creating the study instrument, interpreting data, and reviewing and revising the manuscript. All authors approved the final manuscript as submitted and agree to be accountable for all aspects of the work.

Ethics Declaration

The Children's Hospital of Philadelphia Institutional Review Board deemed the study exempt from the requirement for approval.

K.T.W.) using thematic analysis.[3] The study was deemed exempt by the CHOP Institutional Review Board.

RESULTS

551 neonatologists representing 183 institutions returned complete questionnaires. This represents a response rate of 40% (18/45) for the CHNC, 21% (533/2550) for the SONPM, and 21% overall. 112 participants (73% from level IV NICUs) responded to the free response prompt.

Neonatologists vary on how they use genetic testing in practice (Table 1). Most neonatologists estimate that they send some type of genetic testing on 5–25% of patients. A minority of respondents (19% of the overall cohort) report that they lack on-site clinical geneticists. Few respondents (17%) use standardized institutional criteria for which patients undergo genetic sequencing tests and about half report that they require approval from geneticists before sending sequencing tests. Rapid whole exome sequencing is available to the majority (63%). Most respondents report that neonatologists discuss genetic sequencing tests with parents before sending it (81%) and are responsible for disclosing results (95%). Fewer report that the genetics team talks to families before sending sequencing tests (58%) or that geneticists (81%) or genetic counselors (61%) are responsible for disclosing results.

In free responses, common reasons to embrace testing are (1) clinical utility, (2) increased certainty about decisions, and (3) future utility of increased genetic knowledge (Appendix 1). Reasons for caution around testing include (1) inadequate genetic resources, (2) disutility/delay in care, and (3) possible biases against patients with genetic conditions and risks. Overall, respondents focus on whether genetic results facilitate prediction of long-term outcomes, often beyond NICU discharge and with an emphasis on neurodevelopmental prognosis. Many also emphasize that genetic results should be integrated with family values in making serious medical decisions.

DISCUSSION

Based on the self-report of neonatologists, many patients undergo genetic testing in the NICU today, at times without the support of a genetics team. Neonatologists play a primary role in consenting for and disclosing results of genetic tests. However, physicians outside genetics are not trained to weigh risks and benefits of testing or seek informed consent independently.[4] This is particularly true for complex genetic tests such as exome sequencing, reported here to be widely available. NICUs may be pressured to provide rapid WES and other advanced genetic technologies by the general promotion of genetics within medicine and the public’s increasing awareness of genetic technologies. However, qualitative results reveal neonatologists’ discomfort in using genetic tests with inadequate supports from a trained genetics team, which is consistent with prior work.[2] The national shortage of geneticists and genetic counselors means that NICUs need novel ways to support clinicians as they use genetic tests. Possibilities suggested elsewhere include partnering with large academic centers, telemedicine, improved genetics education, and decision aids for neonatologists.[5] The benefits and risks of sending complex genetic testing must be

constantly evaluated. If neonatologists are inadequately supported in using genetic testing, they may misapply findings.[2]

This study has two limitations that require consideration. First, individual responses may not represent how clinicians practice in all cases or institutional policies. This may also be a strength in that our results capture the ambiguity of clinical practice. Second, the response rate (21%) limits generalizability. We selected listservs to intentionally oversample from level IV NICUs and may have additionally oversampled large NICUs by allowing multiple respondents per NICU (mean 3, maximum 5). Thus, our responses are likely biased toward those working in large, high level NICUs who use genetic test results regularly and may therefore overestimate available genetic supports.

Genetic testing is prevalent and likely to expand in modern NICUs. Tracking how testing is used and whether neonatologists have appropriate support to integrate this technology into clinical practice merits continued attention. Future work should seek to improve neonatologists' comfort and competence in using genetic information and collaborating with genetics services.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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Conflict of Interests

Dr. Joffe received research funding from Pfizer through the University of Pennsylvania until May 2020. Drs. Callahan, Flibotte, Skraban, Wild, Munson, and Feudtner declare no potential competing interests.

Data Availability

All data are available upon request.

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Table 1.

Participant characteristics and responses

Participant Characteristics (N=551)				
Gender	*Female*	62%		
	Male	38%		
NICU Level	*Level II*	3%		
	Level III	37%		
	Level IV	61%		
Testing Practices		**Overall (N=551)**	**Level II/III NICU (N=217)**	**Level IV NICU (N=334)**
Percentage of patients undergoing genetic testing	*< 5%*	19%	39%	6%
	5–10%	35%	35%	35%
	11–25%	30%	19%	37%
	26–50%	13%	6%	18%
	>50%	3%	2%	4%
Clinical geneticist at institution	*No*	19%	41%	5%
	Yes	81%	59%	96%
Standardized criteria for which patients undergo genetic testing	*No*	71%	77%	66%
	Yes	17%	11%	21%
	Not sure	13%	12%	13%
Approval from genetics for sending some types of genetic tests	*No*	46%	54%	41%
	Yes	45%	38%	50%
	Not sure	9%	8%	9%
Ability to send rapid whole exome sequencing (return of results <1 month)	*No*	16%	22%	12%
	Yes	63%	51%	71%
	Not sure	21%	27%	17%
Counseling Practices		**Overall (N=551)**	**Level II/III NICU (N=217)**	**Level IV NICU (N=334)**
What steps do you take to educate a family and seek consent before sending genetic sequencing tests?				
Genetics team usually meets with the family before sending the test		58%	43%	68%
Neonatology team usually discusses the test with the family		81%	87%	78%
Provide written information about genetic testing		29%	34%	26%
Get written consent from the family before sending the testing		30%	4%	33%
In your institution, are medical providers from the following disciplines responsible for disclosing genetic results to parents?				
Neonatologists		95%	98%	93%
Geneticists		81%	64%	92%
Genetic Counselors		61%	45%	71%
Neurologists		48%	43%	51%
Other		10%	9%	10%