SHORT COMMUNICATION

Neonatologists' perspectives on genetic testing procedures

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ABSTRACT

Genetic diseases are common in the neonatal intensive care unit and it has been proven that discovering or confirming these diagnoses has an influence on treatment. However, the availability and usage of genetic testing, particularly exome or genome sequencing, vary greatly amongst NICUs. As a result, we attempted to detect and quantify any variations in practice patterns linked to genetic testing in NICUs across the country. We created a survey that was sent to neonatologists through email. The poll asked about test availability and attractiveness, the procedure of obtaining tests in the NICU, and overall comfort with ordering and interpreting genetic testing. Demographic information on survey participants, as well as information about their NICU, was acquired. The poll was completed by 162 neonatologists from 40 states and 112 different NICUs. Although virtually all (93.2 %) neonatologists rated discovering a genetic diagnosis for their patients as very important, genetic consults were only accessible in 78% of NICUs, and exome or genome sequencing was not provided on a regular basis (69% of NICUs).

Although the majority of US neonatologists questioned believe that genetic studies are necessary for their patients, these tests are not always clinically available. More study regarding implementation difficulties is needed.

Key Words: *Genetic diseases; Epigenetics; Neonatologists*

INTRODUCTION

Many genetic disorders are found in the Neonatal Intensive Care Unit (NICU), where they significantly contribute to admissions, morbidity, and death. Identifying a genetic diagnosis offers several potential advantages, including modifications in medical care, aid with end-of-life decision-making, and family counselling about recurrence risk and prognosis. Indeed, it has been proposed that the NICU is one of the places where diagnostic genetic testing has the biggest influence. Currently, a variety of tests are utilized to determine these illnesses, ranging from chromosomal testing such as karyotyping and Chromosomal Microarray (CMA) to sequencing studies, including gene panels concentrating on a specific indication. More recently, comprehensive genomic examination with gene panels containing hundreds of genes, as well as exome sequencing, have been performed or Genome Sequencing (GS) has been available, and it has been demonstrated to be high yielding and clinically effective. However, genome-wide sequencing assays such as ES/GS are more expensive than more standard testing methods. They are also best delivered with pre- and post-test counselling from a medical genetics practitioner, which is not always accessible in all institutions. As a result, despite the fact that the clinical value and cost effectiveness of genetic testing in the NICU are becoming acknowledged, many NICUs continue to delay it to the outpatient setting. Although the availability of diagnostic genetic testing in NICUs varies greatly, it is constantly developing and has not been measured earlier. Identifying existing practice patterns and heterogeneity in diagnostic genetic testing procedures in NICUs would aid in the development of evidence-based practice recommendations for the genetic evaluation of critically sick newborns and the identification of possible areas for improvement. To obtain insight into these concerns, we polled practicing neonatologists across the United States [1,2].

MATERIAL

We created a poll that addressed many categories about the desirability and availability of genetic tests in the NICU We created the survey questions relating to routinely used genetic tests in the NICU and typical ordering patterns with cooperation from practicing clinical geneticists and neonatologists because no relevant validated instruments were available to collect this information. Following the development of the survey instrument, cognitive interviews with neonatologists and genetic

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counsellors were conducted to strengthen the face validity of our survey items. Supplement 1 contains the whole survey. The poll was distributed through email to over 4000 professionals on the Section on Neonatal and Perinatal Medicine listserv. 14 survey responses were gathered using Research Electronic Data Capture (Vanderbilt University) housed at our university. Participants completed the poll anonymously, albeit they were asked to name the NICU in which they work. For the examination of practice patterns associated to a NICU (rather than an individual neonatologist), we limited answers to unique NICUs only when two or more replies were obtained from the same location. For completed surveys in which the participant did not disclose the NICU name, survey replies reflecting data at the NICU level were included if other factors permitted us to identify it as a separate NICU. For our NICU level studies, responses were pooled such that each NICU was only represented once. If responders from the same NICU gave different replies to questions about test availability (to which a single response was sought), the response that indicated that the test was available was utilized. Otherwise, if two replies from the same NICU were divergent and could not be aggregated (i.e., responses were mutually exclusive), the question for that NICU was left unanswered. When it came to turnaround time, the most frequent response was used if numerous choices were picked from the same NICU or an average of divergent replies if there was no majority [3-5].

Overall, 21 NICUs had >1 respondent for the same NICU,sand of these, the replies differed the most for questions spertaining to the procedure of transmitting sequencing-based tests, although multiple responses were allowed for these questions. SPSS version 27 (IBM Corporation) was utilized for analysis, with Fisher exact test or 2 test employed to compare categorical data. The Boston Children's Hospital Institutional Review Board authorized this study, and completion of the survey constituted informed consent [6].

RESULT

From April to July 2021, 162 neonatologists completed the study survey, with a response rate of about 4%. A total of 40 states and Washington DC were represented (missing: Alaska, Idaho, Maine, New Hampshire, North Dakota, Rhode Island, South Dakota, Vermont, West Virginia, and Wyoming), with replies ranging from 1 to 11 from different NICUs. Two responders were from outside the United States, one from Puerto Rico, and three did not offer the state or NICU name. Other demographic characteristics are mentioned.

DISCUSSION

We give a summary of current genetic testing practice patterns in NICUs as stated by neonatologists, as well as their perspectives on genetic testing. Overall, we discovered that most neonatologists thought a genetic diagnosis was crucial, however a sizable number saw a disparity between demand and availability, particularly for ES/GS.

Established tests, like as karyotyping and CMA, are simpler for the NICU team to request independently, as predicted, but newer, sequencing-based assays often require monitoring to guarantee optimal usage and assist in test interpretation and counselling. These findings are consistent with previous research on patient satisfaction with genomic sequencing and previous studies of GS for critically sick newborns, which revealed that NICU clinicians valued the information, acquired from such testing and utilized it to direct medical treatment. 16 We also discovered that genetic diagnoses are highly desired by neonatologists; yet, the tests required to make a diagnosis are not now regularly accessible. Interestingly,

neonatologists who were older and had been in practice for a longer period of time indicated a greater perceived value of genetic testing, which might be influenced by past experience with genetic testing. Our findings point to a greater need for clinical genetics engagement in NICU settings, where test availability has lagged behind current research. Because karyotyping and CMA are unlikely to detect all babies with uncommon genetic abnormalities. Because the diagnostic and clinical value of ES/GS in this group is well known, 1 more research into the introduction of these diagnostic techniques into the NICU context is desirable, particularly in settings where clinical genetics teams are not accessible.

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