Supporting Information: Perinatal Exome Sequencing Survey administered to survey respondents. Downloaded directly from REDCap. Includes coversheet, demographic information questions, and survey questions in a PDF format.

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Dear Potential Research Subject:

You are invited to participate in a research study investigating the medical community's opinion of perinatal exome sequencing (ES). The Molecular and Human Genetics Department at the Baylor College of Medicine is surveying English speaking adults (18+) who are a part of the local and national medical community. This population includes, but is not limited to: midwives, genetic counseling students, genetic counselors, laboratory scientists, medical students, obstetric gynecology and/or genetics residents, attendings, and clinical/post-doctoral fellows.

Exome sequencing is predicted to be utilized as a prevalent method for perinatal diagnosis in years to come. Prior to the wide employment of perinatal ES, it is essential to assess the medical community's opinion on how it should be clinically implemented. The study will explore the local and national medical community's attitude towards the use of ES in perinatal medicine. The information you provide will help us better understand the medical community's opinion on this topic.

Participation in this study involves the completion of a survey that asks you basic questions about yourself and how you feel about perinatal exome sequencing. This survey is anonymous and we will not collect any identifiable information. The survey should take about 15 minutes or less to complete. By filling out this survey and submitting it, you are providing consent to be in this study.

There are no physical risks in participating in the research; however, there is a small risk for the loss of confidentiality. The study personnel will make every effort to minimize the risks by not collecting any identifiable health information and keeping your survey anonymous. Your choice to participate in this study will have no effect on your rights or benefits. There are no costs or monetary payments if you decide to take part in this study.

If you have any questions about this survey or the study, please contact Kylie Johnson at Kylie.Johnson@bcm.edu. If you have additional questions about your rights as a research subject, contact the Institutional Review Board for Human Subject Research for Baylor College of Medicine & Affiliated Hospitals at (713) 798-6970.

Thank you for your time.

Sincerely,

Kylie Johnson, M.D. candidate, Baylor College of Medicine
Salma Nassef, M.S. C.G.C., Baylor College of Medicine
Ignatia Van den Veyver, M.D., Baylor College of Medicine

Demographic Questions	
To which gender do you most identify?	◯ Female ◯ Male ◯ Transgender female ◯ Transgender male ◯ Gender variant/non-conforming ◯ Prefer not to answer
What profession are you trained/training in?	◯ Genetic counseling ◯ PhD scientist ◯ Medicine ◯ Midwifery
Select the option that best reflects your level of training:	◯ Student ◯ Practicing genetic counselor
Select the option that best reflects your level of training:	◯ Trainee ◯ Researcher ◯ Lab director ◯ Lab fellow ◯ Post-doctoral lab fellow
Select the option that best reflects your level of medical training:	◯ Medical student ◯ Intern/Resident ◯ Clinical/post-doctoral Fellow ◯ Attending
In which specialty are you an Intern/Resident?	◯ Obstetrics and gynecology ◯ Genetics ◯ Other
In which specialty are you a Fellow?	◯ Maternal fetal medicine ◯ Genetics ◯ Maternal fetal medicine + genetics combined ◯ REI ◯ Other
In which specialty are you an attending?	◯ Obstetrics and gynecology ◯ Genetics ◯ Other
How many years have passed since your last full-time post graduate training?	◯ Still in training ◯ 0-5 ◯ 6-10 ◯ 11-15 ◯ 16-20 ◯ 21+ (Round down to the greatest whole number)

Survey Questions

Instructions: Please answer all of the following questions to the best of your ability. If you do not know the answer to a question, please opt to select either the "I do not know" or "Prefer not to answer" answer choice.

Do you know what Exome Sequencing is?

- ◯ Yes
- ◯ Somewhat
- ◯ No

Please read the following definition:

Exome sequencing (ES) studies the coding regions of DNA otherwise known as exomes. Exomes encompass greater than 85% of known pathogenic variants. ES elucidates the DNA sequence of these coding regions base by base with high resolution.

Have you read/are able to recall any published guidelines for the use of Exome sequencing in a perinatal clinical setting?

- ◯ Yes
- ◯ No

Please elaborate on the previous question:

How do you feel about the following statements?	Strongly agree	Moderately agree	Neutral	Moderately disagree	Strongly disagree	Prefer not to answer
Everyone should receive ES at birth.	○	○	○	○	○	○
Prenatal ES should never be used.	○	○	○	○	○	○
Invasive prenatal testing (chorionic villus sampling or amniocentesis) should be offered to every pregnant woman.	○	○	○	○	○	○
Prospective parents should be able to request prenatal ES without medical indication.	○	○	○	○	○	○
Prenatal ES should be the first diagnostic genetic test ordered in the context of an abnormal mid-pregnancy ultrasound.	○	○	○	○	○	○
Prenatal ES should be the genetic test ordered in the context of an abnormal mid-pregnancy ultrasound BUT only after karyotyping + chromosomal microarray tests fails to identify an underlying genetic etiology.	○	○	○	○	○	○
Prenatal ES should be the first diagnostic genetic test ordered in the context of a positive combined first screen (including information from ultrasound and maternal serum analysis) for trisomy 21.	○	○	○	○	○	○
Prenatal ES should always be used in the context of advanced maternal age (35 years +).	○	○	○	○	○	○
Prenatal ES should be offered to every prospective mother, irrespective of risk, in her first trimester.	○	○	○	○	○	○
Prenatal ES should be offered to every prospective mother, irrespective of risk, in her second trimester.	○	○	○	○	○	○

ES should be performed on all stillbirths.	◯	◯	◯	◯	◯	◯

A 25-year-old woman in her first pregnancy presents to your clinic at 19 weeks gestational age for her mid-pregnancy ultrasound. Fetal anomalies are detected on the ultrasound and the woman agrees to an amniocentesis. What is the best genetic test to order?

- ◯ Karyotyping
- ◯ Chromosome microarray analysis
- ◯ Chromosome microarray analysis + Karyotyping
- ◯ Genome wide sequencing
- ◯ Gene specific sequencing
- ◯ I do not know

If fetal anomalies are detected on the mid-pregnancy ultrasound AND karyotyping/ chromosome microarray tests fail to identify an underlying genetic etiology, which genetic test would you next choose to order?

- ◯ Single gene test
- ◯ Gene panel
- ◯ Exome sequencing
- ◯ Genome sequencing
- ◯ I do not know
- ◯ None of the above

A 27-year-old woman in her third pregnancy presents to your clinic at 18 weeks gestational age for her mid-pregnancy ultrasound. Fetal anomalies are detected on the ultrasound and the woman agrees to an amniocentesis. Subsequent karyotyping and chromosomal microarray tests are normal, and you want to offer ES. When is the best time to achieve patient consent for prenatal ES?

- ◯ During the initial counseling for amniocentesis
- ◯ After G-banded karyotyping and chromosomal microarray tests fail to identify the underlying genetic etiology
- ◯ I do not know

What is the first test you would order in the context of a stillbirth?

- ◯ G-banded karyotyping
- ◯ Chromosome microarray analysis
- ◯ G-banded karyotyping + Chromosome microarray analysis
- ◯ None of the above
- ◯ I do not know

If chromosomal microarray analysis results are normal in the context of a stillbirth what is the next immediate diagnostic test you would choose to order?

- ◯ Exome sequencing
- ◯ Genome sequencing
- ◯ None of the above
- ◯ I do not know