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Better fetal diagnosis and
through next-generation

Next generation sequencing (NGS) is one area of precision medicine that holds promise for the diagnosis and treatment of fetal congenital anomalies, genetic disorders and other complications of pregnancy. This technology allows UCSF clinicians and researchers to examine the exome and to better understand and treat fetal conditions.

The UCSF Center for Maternal-Fetal Precision Medicine is leading new advances in gene-based interventions for fetal care. Two clinical trials of exome sequencing are now underway through the Center for Maternal-Fetal Precision Medicine at UCSF Benioff Children's Hospitals.

Benefits of exome sequencing

- Thousands of genes can be analyzed at the same time, rather than testing one or a few genes at a time.
- Exome sequencing may identify or rule out specific treatments for a patient's individual diagnosis (though exome sequencing does not always produce actionable findings).
- Results can provide information about the chance of recurrence of the same condition in another pregnancy, and allow for more accurate prenatal diagnosis in the future.

What is exome sequencing?

More efficient than tests that look at single genes one at a time, exome sequencing is a genomic technique for analyzing all the protein-coding regions of the genome. These regions are referred to as the exome. Focusing on this small portion (only 2 percent) is less expensive than sequencing the whole genome but still identifies a genetic diagnosis in many cases.

The goal of exome sequencing is to cast a wider net than is possible with specific gene panels, to more quickly identify genetic etiologies of diseases. Researchers at UCSF Benioff Children's Hospitals are using exome sequencing to better understand the causes of fetal anomalies. The result may improve patient care.

Webinar: When Is Exome Sequencing Valuable in Prenatal Diagnosis?

View the webinar ^[1], with maternal-fetal medicine specialist, Mary E. Norton, MD, discussing genetic testing options, with a focus on when exome sequencing has value to families. Also, perinatologist, Teresa N. Sparks, MD, presents specifics on exome sequencing for nonimmune hydrops fetalis, caused by many single-gene disorders, plus a look at findings from her current NIHF study.

Prenatal exome sequencing clinical trial now enrolling patients

The Center for Maternal-Fetal Precision Medicine at UCSF Benioff Children's Hospitals is currently enrolling for two clinical trials performing exome sequencing. The goals of these studies include:

- To determine whether exome sequencing is a useful tool for prenatal diagnosis;

- To define the genetic contributions to non-immune hydrops fetalis (NIHF); and
- To develop a precision-based approach to prenatal diagnosis and management, ultimately with targeted treatments specific to the genetic cause of disease.

Pregnant women who meet the following criteria may be eligible:

- An ultrasound has identified a fetal structural anomaly (or multiple anomalies) in a major organ system.
- Prenatal testing done through amniocentesis or chorionic villus sampling (CVS) has not identified the cause of the structural anomaly.
- At least one biological parent is available to provide a DNA sample (blood or saliva) for completion of the exome sequencing test.

All costs of testing will be paid for by these studies, which are funded by the National Human Genome Research Institute of the National Institutes of Health.

For a full description of these studies:

Download the PEGS Research PDF Brochure^[2]

or visit UCSF Clinical Trials to read the specific trial information at:

<https://clinicaltrials.ucsf.edu/trial/NCT03482141> ^[3]

<https://clinicaltrials.ucsf.edu/trial/NCT03412760> ^[4]

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Links

[1] <https://medconnection.ucsfhealth.org/videos/genetic-testing-update-when-is-exome-sequencing-valuable-in-prenatal-diagnosis>

[2] https://fetus.ucsf.edu/sites/fetus.ucsf.edu/files/wysiwyg/ENG%20PEGS%20Prenatal%20Brochure_02.05.2018%20F

[3] <https://clinicaltrials.ucsf.edu/trial/NCT03482141>

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