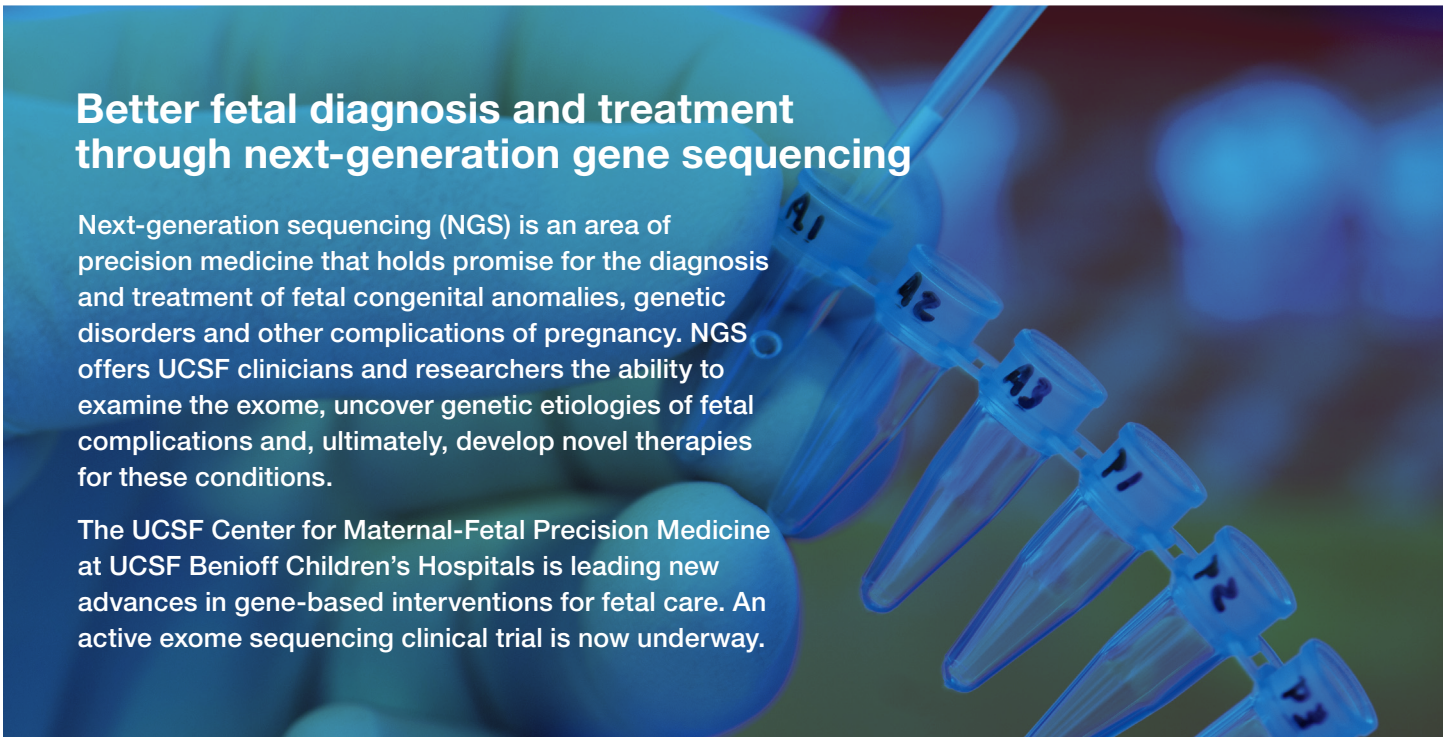
UCSF Benioff Children's Hospitals
Fetal Treatment Center

Exome Sequencing

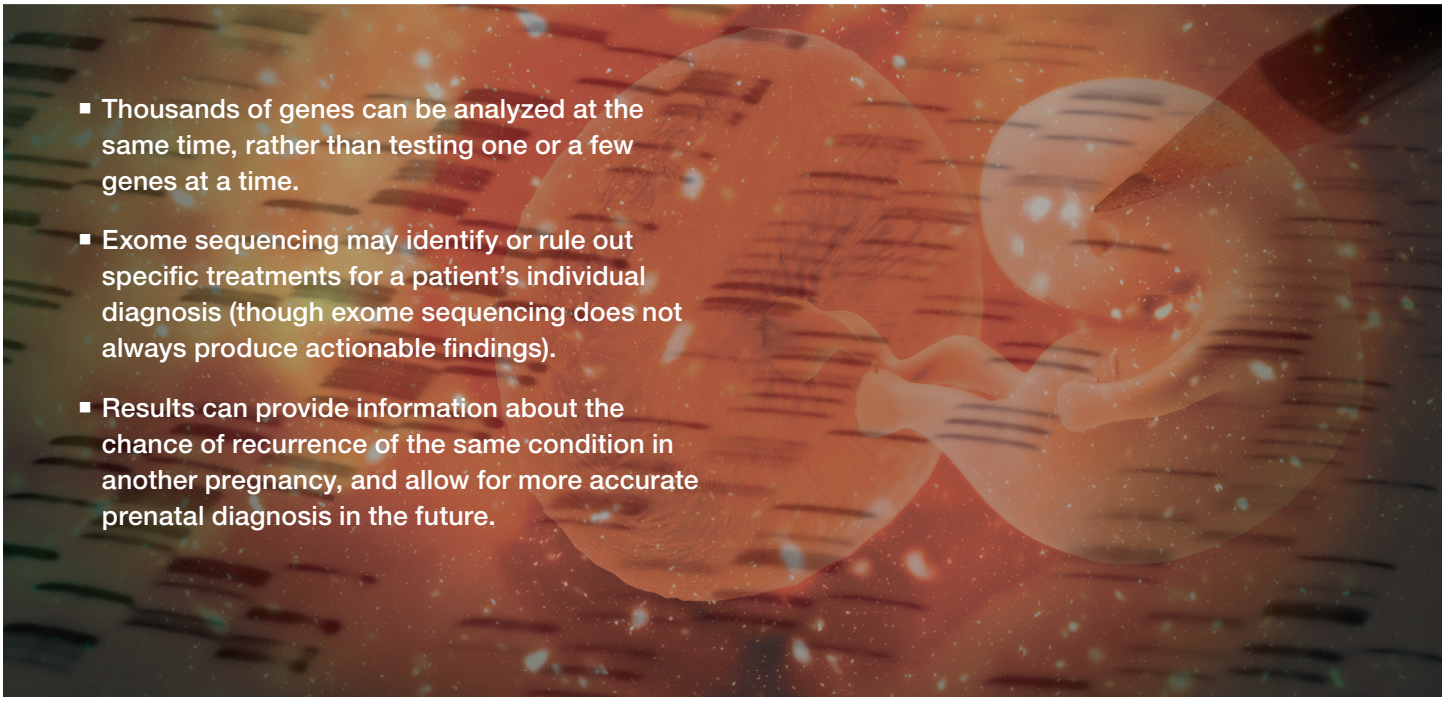


Better fetal diagnosis and treatment through next-generation gene sequencing

Next-generation sequencing (NGS) is an area of precision medicine that holds promise for the diagnosis and treatment of fetal congenital anomalies, genetic disorders and other complications of pregnancy. NGS offers UCSF clinicians and researchers the ability to examine the exome, uncover genetic etiologies of fetal complications and, ultimately, develop novel therapies for these conditions.

The UCSF Center for Maternal-Fetal Precision Medicine at UCSF Benioff Children's Hospitals is leading new advances in gene-based interventions for fetal care. An active exome sequencing clinical trial is now underway.

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.

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What is exome sequencing?

More efficient than tests that look at 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.

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

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.

We are now able to offer exome sequencing to all eligible patients as part of our comprehensive care. Most cases of fetal physical anomalies are eligible.

Our Team

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