Our goals are to develop a precision-based approach to care for NIHF, and ultimately, targeted treatments specific to each underlying cause of NIHF.

For more information or to refer a patient, please contact us:

UCSF Fetal Treatment Center

**HyDROPS Team**

1-800-RX-FETUS
(1-800-793-3887)

hydrops@ucsf.edu

Teresa Sparks, MD, MAS
Mary Norton, MD
Billie Lianoglou, LCGC
Tippi MacKenzie, MD
Stephan Sanders, MD, PhD
Renata Gallagher, MD

Our research contributes novel information about genetic disorders that cause Non-Immune Hydrops Fetalis. This enables more accurate counseling, individualized antenatal care, and anticipation of neonatal needs.
**STUDY OVERVIEW**

- Diagnosis of NIHF
- Standard work up including amniocentesis or CVS during pregnancy, or diagnostic testing after birth
- Contact UCSF here or at any earlier time
- Samples to UCSF for trio exome sequencing (fetus or infant, and both biological parents)
- UCSF team returns results to the participant and referring provider

**STUDY DESIGN**

- **Enrollment**
  - The UCSF team can consent and enroll patients locally at UCSF or remotely via video or telephone.
- **Inclusion Criteria**
  - Living or demised fetus or infant with non-diagnostic karyotype and/or microarray, and one or more of the following:
    - Non-immune hydrops fetalis
    - Single abnormal fetal fluid compartment (such as isolated ascites)
    - Cystic hygroma
    - Nuchal translucency of \( \geq 3.5 \) mm
- **Exclusion Criteria** (any)
  - Alloimmunization
  - Twin-twin transfusion syndrome
- **Approach to Testing**
  - Trio exome sequencing on fetus/infant and (ideally) both biological parents
  - If patient declines diagnostic testing during pregnancy, infant or other tissue sample may be sent to UCSF
  - Exome sequencing at no cost to families
  - Multidisciplinary panel review and classification of genetic variants
  - Turnaround time is 2 - 4 weeks for ongoing pregnancies and live births
  - Our CLIA-approved lab issues a formal report to the patient and ordering provider

**HOW TO PARTICIPATE**

- **Clinical Data**
  - Our team will request a limited amount of clinical information, such as results of karyotype and/or microarray and ultrasound findings.
- **How to Refer a Patient**
  - hydrops@ucsf.edu or 1-800-RX-FETUS
  - See insert for recommended work up of NIHF.
- **Send Us a Specimen**
  - Our team can assist with sample transfer
  - Saliva kits shipped directly to biological parents with pre-paid return to UCSF
  - Only one sample type needed for the fetus/infant as well as each biological parent
  - Fetal: Cultured amniocytes, chorionic villi, cord blood, products of conception, extracted DNA, or other tissue
  - Neonatal: Blood, buccal/saliva, skin, or other tissue
  - Parental: Blood or saliva
### Work up of Non-Immune Hydrops Fetalis (NIHF)

*Adapted from SMFM Clinical Guideline, 2015*

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<th>Ultrasound including fetal echocardiogram &amp; MCA Dopplers; type and screen; MCV*; KB</th>
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<td>Fetal anemia</td>
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**NO**

- Amniocentesis

- **Send for:**
  - Karyotype and/or microarray;
  - +/-PCR for CMV, toxoplasmosis, parvovirus

  - If karyotype and/or microarray are non-diagnostic, consider:
    - RASopathy panel
    - Other targeted testing if high clinical suspicion
    - Exome sequencing

**YES**

- Amniocentesis or fetal blood sample if IUT

- **Send for:**
  - Karyotype and/or microarray;
  - PCR for CMV, toxoplasmosis, parvovirus

  - If evaluations are non-diagnostic, consider:
    - Targeted testing for hereditary anemias if clinical suspicion
    - Exome sequencing

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*Low MCV: test for alpha thalassemia*

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