PEARL (PrEnAtal Enzyme Replacement Therapy for Lysosomal Storage Disorders)

NCT04532047

Status	RECRUITING
Phase	Phase 1
Sponsor	University of California, San Francisco
Enrollment	10 participants

Key Eligibility Criteria

Inclusion (5)

- Live male or female fetuses at 18 0/7 weeks to 34 6/7 weeks gestation
- Diagnosis of one of the 8 included LSDs in utero by genetic or enzymatic analyses performed on amniotic fluid, fetal blood, placental tissue, or other samples through chorionic villus sampling (CVS), amniocentesis, cordocentesis, cell free fetal DNA, or other procedures. In the event that parents are identified as genetic carriers for a LSD, diagnostic testing for the fetus would be performed to confirm the diagnosis
- Pregnant women age 18 years to 50 years, carrying a live male or female fetus at 18 0/7 weeks to 34 6/7 weeks gestation
- Identified through the above listed means to be carrying a fetus with an LSD.
- Ability to give written informed consent and comply with the requirements of the study.

Exclusion (13)

- Fetuses with a concurrent severe structural anomaly
- Fetuses with an additional pathogenic genetic variant not related to the underlying LSD that contribute a significant risk of morbidity or mortality.
- Hydrops fetalis will not be an exclusion criterion because ERT has the possibility of significant benefit in this situation.
- Women with one or more significant comorbidities that would preclude fetal intervention including, but not limited to:
- inability to complete the procedure secondary to maternal body habitus or placental location

... and 8 more (see full listing online)

Locations (1 total)

University of California, San Francisco, California, United States