

PHASE 1 CLINICAL TRIAL OF IN UTERO ENZYME REPLACEMENT THERAPY

PEARL Trial—PrEnAtal enzyme Replacement for Lysosomal storage diseases.

New PEARL Trial website: <https://pearltrial.ucsf.edu>.

The aim of the PEARL Trial is to establish the safety and efficacy of prenatal ERT for lysosomal storage diseases, including:

- ☐ **Mucopolysaccharidoses (MPS) I, II, IVa, VI, VII**
- ☐ **Infantile-onset Pompe disease (IOPD)**
- ☐ **Neuronopathic Gaucher disease (types 2 and 3)**
- ☐ **Wolman disease**

We have seen promising results with this approach—the first patient who was treated with in utero enzyme replacement therapy had infantile-onset Pompe disease and we saw a clear benefit of the prenatal therapy on her motor function, as we published in Cohen et al, [NEJM 2022](#).

We have treated several additional patients at UCSF with no apparent safety concerns and have presented promising improvements in biomarkers at national conferences, including WORLD 2024.

Like other rare disease clinical trials, reaching eligible patients is challenging. Some participants will learn about the PEARL Trial through online searches. However, direct referral by clinicians and others in the lysosomal disease community is critical.

Here are two ways you can help:

1. **If you care for families who have previously experienced an affected pregnancy**, please consider sharing this information with them so that they are aware of this option for a future pregnancy.
2. **Forward this message** to individuals who care for patients, diagnose, or study lysosomal diseases.

To refer a patient to the [PEARL Trial](#), please contact our clinical trial manager, Emma.Canepa@ucsf.edu.

We are also keeping a [registry of patients](#) who are prenatally diagnosed with the above conditions so please contact us if you are aware of families who may consider participating.

Thank you for partnering with us to bring this promising therapy to patients.

Sincerely,

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Related links:

[UCSF PEARL Trial](#)

[UCSF Fetal Treatment Center](#)

[Recorded Session on Prenatal ERT](#)