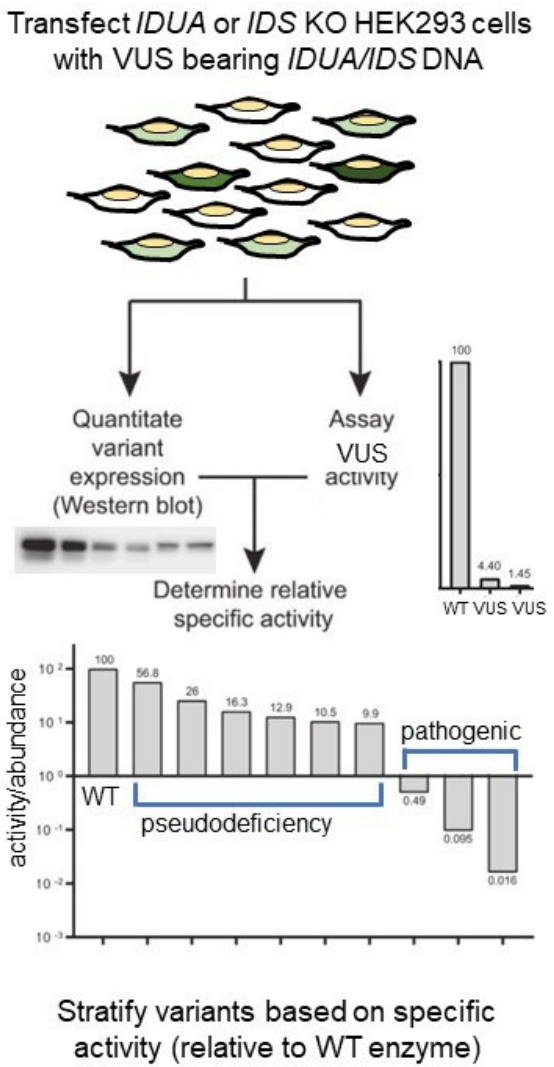
VARIFY

A cell-based platform for functional characterization of *IDUA/IDS* variants

Greenwood Genetic Center
Where Compassion Inspires Progress



ABOUT

The Greenwood Genetic Center has developed a functional biochemical platform for the characterization of individual *IDUA* and *IDS* variants for MPS I and MPS II, respectively.

VARIFY is an investigational analysis to provide additional in vitro functional analysis for individual molecular variants in the IUDA or IDS genes. With increasing access to molecular testing as well as implementation of newborn screening, there is an increased clinical need to have functional evidence for the classification of variants of unknown clinical significance.

This robust cell-based platform relies on transfection of knock out cells with gene-specific DNA containing the variant of interest. A combination of Western blot analysis and enzyme activity assay is used to determine the relative specific activity of wild-type and individual variants. (see figure to left)

SUMBISSION REQUIREMENTS

GGC is interested in offering analysis to characterize additional *IDUA* and *IDS* variants.

- We welcome inquiries or submissions of novel variants of unknown clinical significance in either the *IDUA* or *IDS* genes.
- No patient specimens will be required.
- Biochemical testing for the patient carrying the VUS will be required.
- Each variant will need to be reviewed and approved by GGC.

We also welcome inquiries regarding additional collaborations for other lysosomal diseases.

PUBLICATION

Yu SH, Pollard L, Wood T, Flanagan-Steet H, Steet R. A Biochemical Platform to Define the Relative Specific Activity of IDUA Variants Identified by Newborn Screening. Int J Neonatal Screen. 2020 Nov 12;6(4):88. doi: 10.3390/ijns6040088. PMID: 33198351; PMCID: PMC7711455.

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