CONCLUSION

- Our data set for the three lysosomal storage disorders indicated biochemical results alone are insufficient for variant reclassification albeit it’s one of the critical components.
- Additional information from multiple measurements through multiomics, large reference and patient databases, integrated analysis methods, and computational infrastructure is required to further interpret the VOUS variants to ensure improving early diagnosis and personalized treatment.
- Advanced Omics technologies will help discover of additional biomarkers and further elucidate the phenotype-genotype relationship along with tracking response to new therapies.
- Accurate variant classification and interpretation through the incorporation of omics will help personalized genetic counseling to reduce unnecessary medical visits and family anxiety.

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