Aim 2

- Develop and integrate novel computational tools into the Functional annotation pipeline (SVIM) pipeline to evaluate the impact of SVs by
- identifying genomic elements affected by a variant and the type of impact
- assessing the impact based on the types of SV and disruption mechanism
 - up-weighting SVs associated with certain

functional features

• Using the new pipeline, prioritize SVs from the reference set to identify high-impact variants

