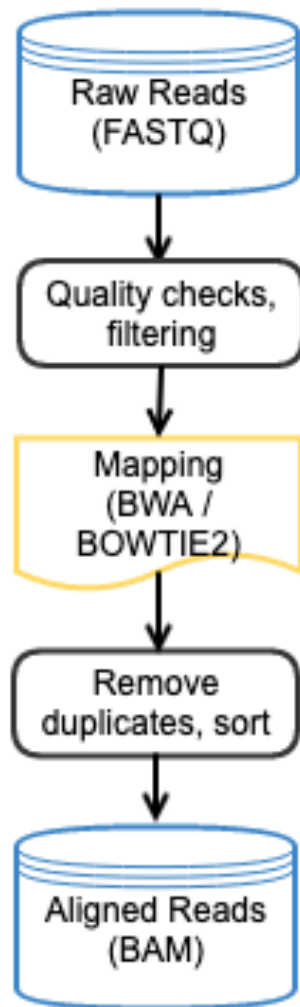
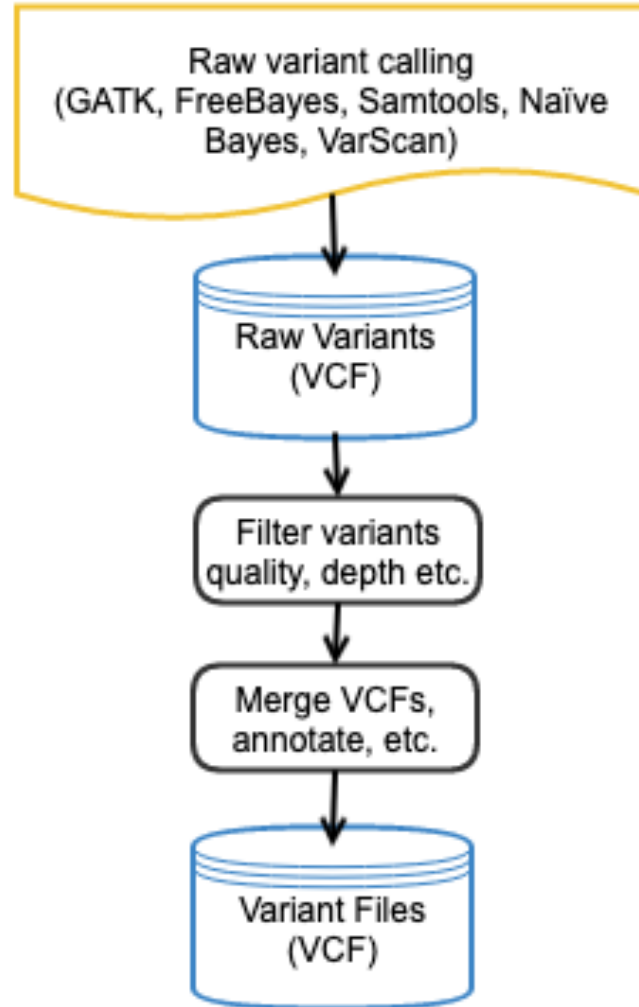


# Background & Workshop Overview

## PREPROCESSING



## VARIANT CALLING

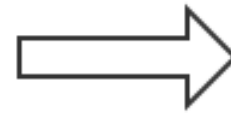
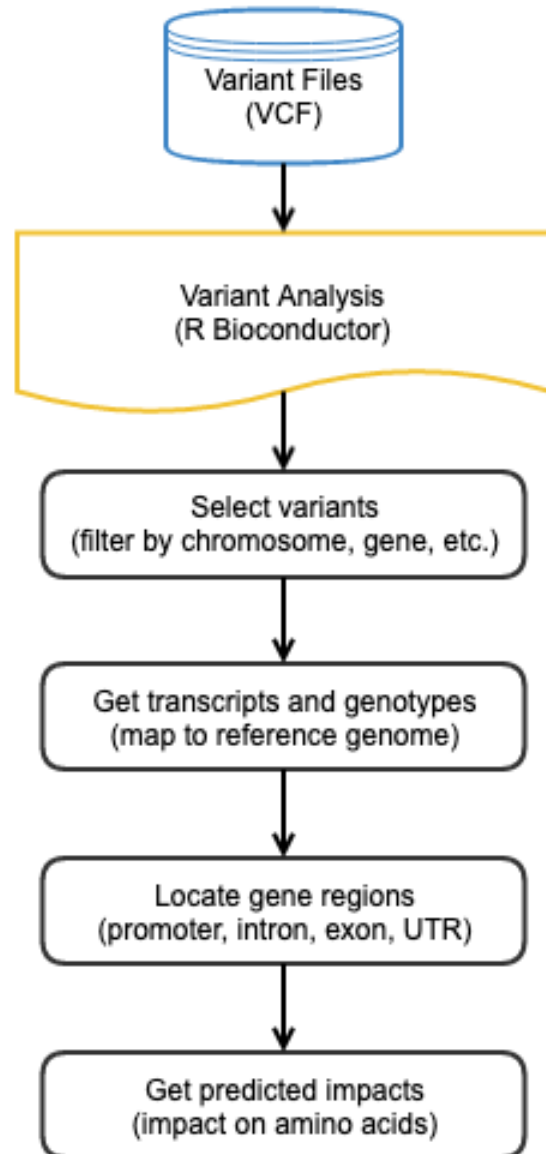


## VARIANT ANALYSIS

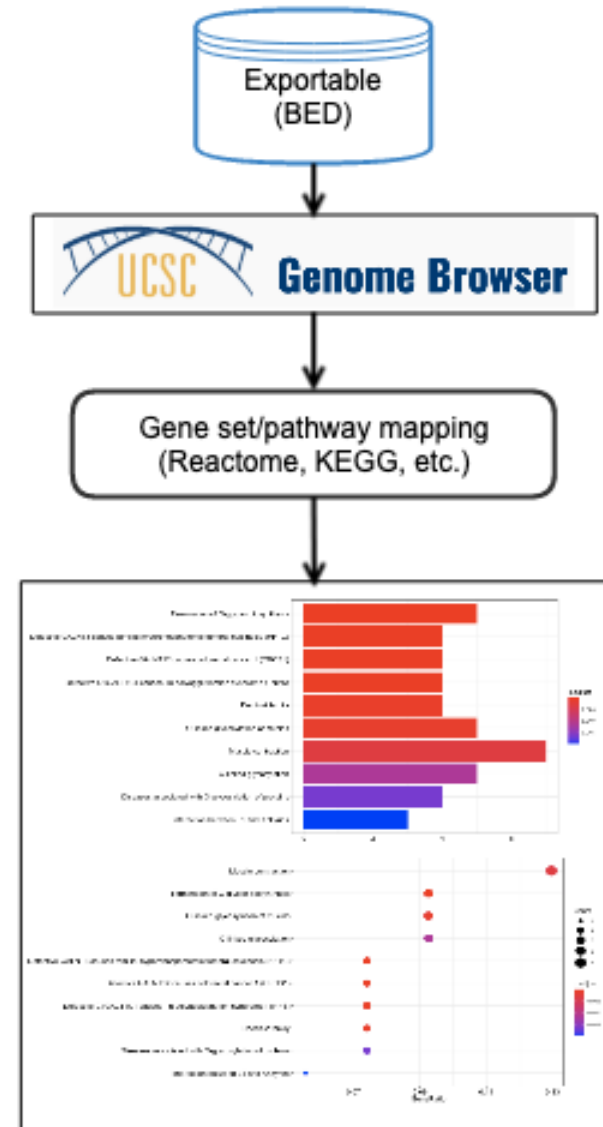


Overview of steps covered in this workshop:

### VARIANT ANALYSIS (FUNCTIONAL IMPACTS)



### VARIANT ANALYSIS (VISUALIZATION)



# Visualizations

## Using R Bioconductor Packages



# UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly

move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x 100x

chr17:3,012,642-4,012,641 1,000,000 bp. enter position, gene symbol, HGVS or search terms go

chr17 (p13.3-p13.2) 13.3 13.2 p13.1 17p12 17p11.2 17q11.2 17q12 21.31 17q22 24.3 q25.1 17q25.3



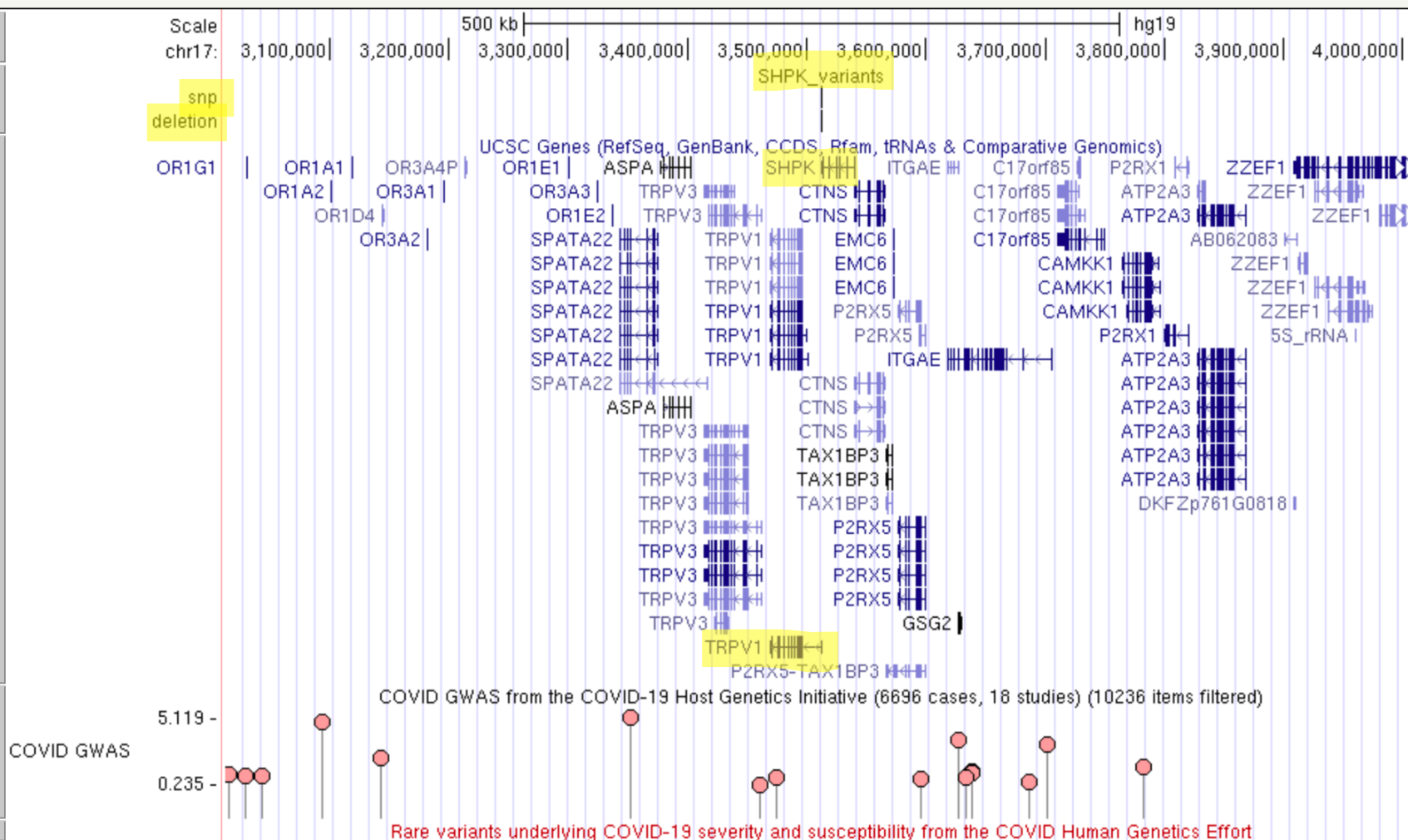
Zoomed-out view of SHPK variants (snp, del)

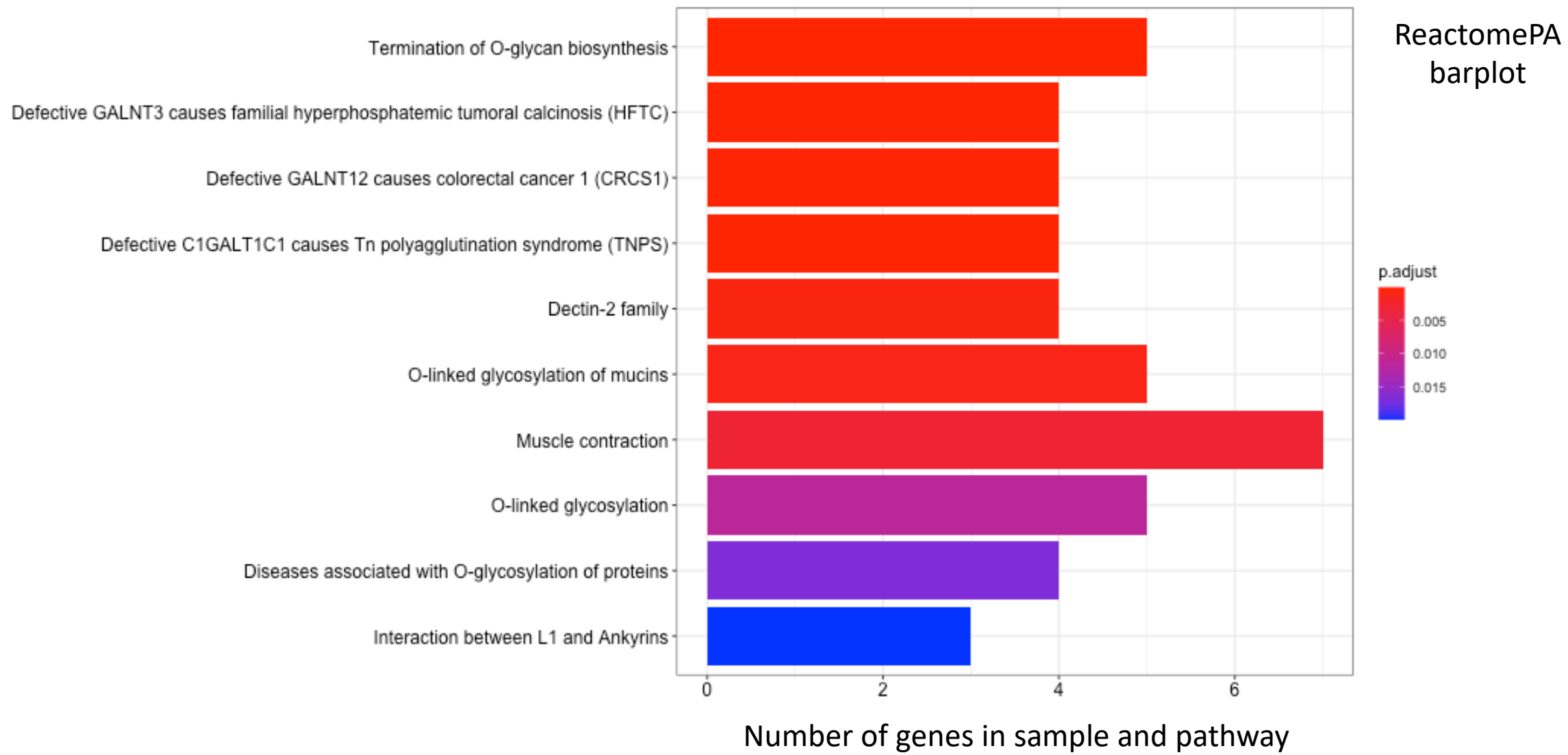
→ SHPK gene

*This is just to show how much fun you can have once you get your sample variants mapped to UCSC Genome Browser (zoom/select tracks)!*

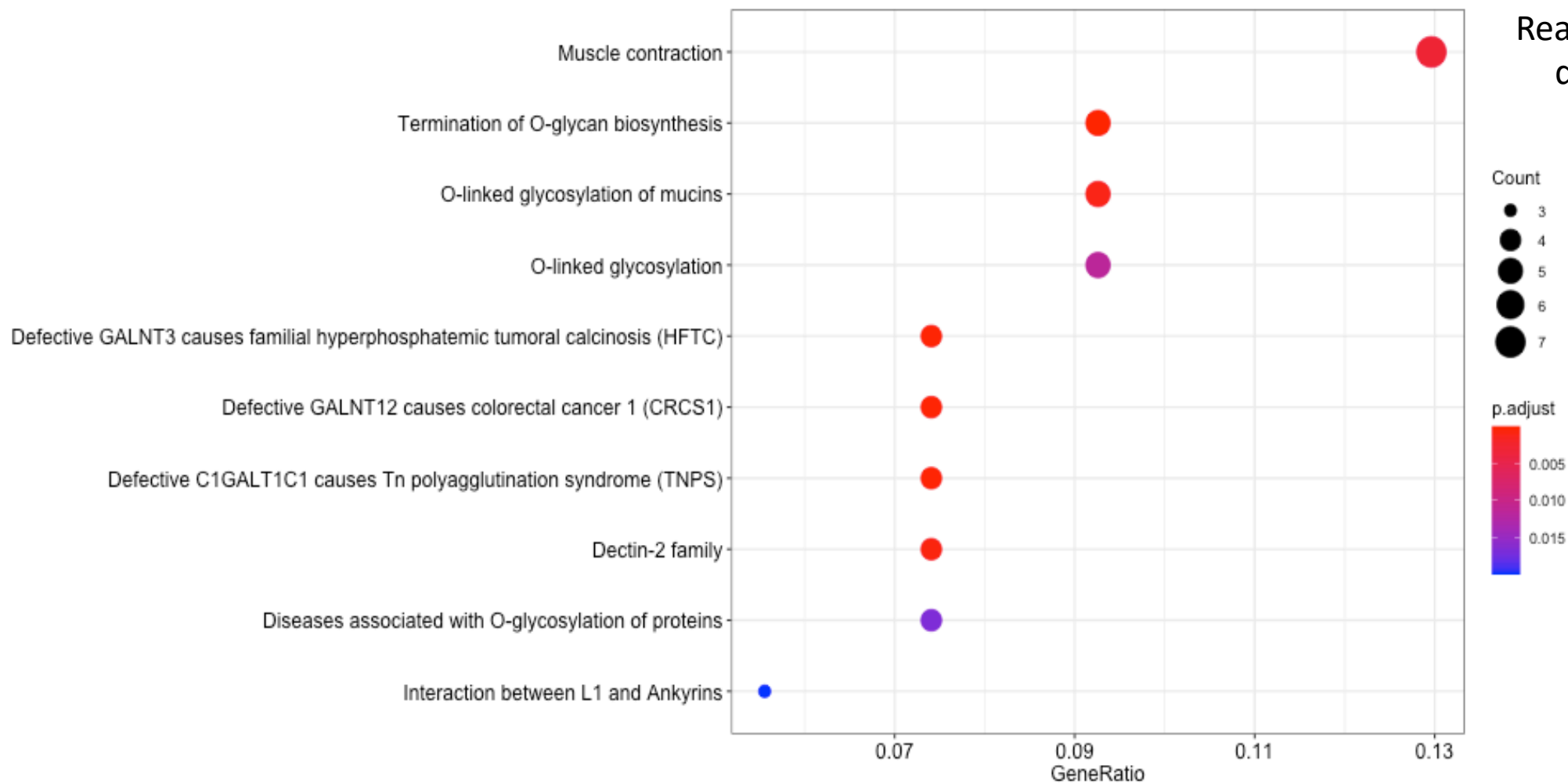
→ TRPV1 gene

New COVID GWAS track





# ReactomePA dotplot





## ReactomePA emapplot

