CNV detection
(with PennCNV software)

CNV evaluation
with omics-based metrics

Modelling omics-informed
CNV quality score (OQS)

OQS model

Prediction of CNV quality
using OQS model
(prior genetic association
analyses)

Datasets with genotyping array intensities and omics layers available

Datasets with genotyping array intensities available

pCNV carrier i

CNV call under evaluation (pCNV)

Overlap

CNV calls from sequencing data

WGS metric = Overlap (bp) / pCNV (bp)

X_i – gene expression/
methylation intensity
for pCNV carrier i

Density

$\Phi(x_i)$

$1 - \Phi(x_i)$

*Gene expression/methylation intensity
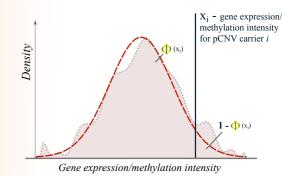across non-carrier samples*

GE/MET metric = $\Phi(x_i) - (1 - \Phi(x_i))$