

Experimental Workflow

Stage 1: CNV discovery

Assay: Agilent aCGH 244k Custom Arrays
(168 ASD cases, 149 Controls)



29 Loci Prioritized

Stage 2: Molecular Confirmation & Additional Population screening For Prioritization

Taqman CNV Assays
(Confirmed CNVs individuals from stage 1, 170 ASD cases, 170 controls)



11 Loci Prioritized for additional screening

Stage 3: Family-based Validation

Taqman CNV Assays
(755 ASD cases, 1809 controls)



2 genes, NRXN1 & GABBR2, passed family-based validation