

Description of Additional Supplementary Files

File Name: Supplementary Data 1

Description: Discovery Cohort Characteristics

File Name: Supplementary Data 2

Description: Summary of WGS quality and number of variants detected

File Name: Supplementary Data 3

Description: Validation status of de novo exonic SNVs and indels in discovery cohort

File Name: Supplementary Data 4

Description: Validation status of de novo exonic CNVs in discovery cohort

File Name: Supplementary Data 5

Description: Clinically significant and ASD candidate SNVs and indels in discovery cohort

File Name: Supplementary Data 6

Description: Clinically significant and ASD candidate CNVs, SVs, and tandem repeat expansions in discovery cohort

File Name: Supplementary Data 7

Description: Phenotype description of 69 probands with clinically significant or ASD-candidate variants in discovery cohort

File Name: Supplementary Data 8

Description: Prevalence of ASD variants in morphologic subtypes of ASD in the discovery cohort

File Name: Supplementary Data 9

Description: Prevalance of SNVs, indels and CNVs in 43 genesets among ASD subtypes in the discovery cohort

File Name: Supplementary Data 10

Description: Prevalance of SNVs, indels and CNVs in noncoding regions among ASD subtypes in the discovery cohort

File Name: Supplementary Data 11

Description: : Coefficients of significant gene sets and regions in the discovery cohort (classified by gold standard dysmorphology) using 10x30-fold cross validation

File Name: Supplementary Data 12

Description: Phenotype, GRVS and PRS information for discovery cohort

File Name: Supplementary Data 13

Description: Average scores for clinically significant variants, all and remaining ASD-relevant variants in the discovery cohort that occur in ASD relevant regions identified by gene set enrichment

File Name: Supplementary Data 14

Description: Coefficients of significant gene sets and regions in the ADM-classified discovery cohort under 10x30-fold cross validation*

File Name: Supplementary Data 15

Description: Coefficients of gene sets and regions with $P < 0.1$ in the ADMclassified discovery cohort

File Name: Supplementary Data 16

Description: Phenotype, GRVS and PRS information for replication cohort

File Name: Supplementary Data 17

Description: Clinically significant and ASD candidate SNVs and indels in the replication cohort

File Name: Supplementary Data 18

Description: Clinically significant and ASD candidate CNVs in replication cohort

File Name: Supplementary Data 19

Description: Variant predictors and databases used for variant annotation

File Name: Supplementary Data 20

Description: noncoding regions examined in this study

File Name: Supplementary Data 21

Description: Gene sets used in gene set enrichment analysis