Figures

Figure 1: Score variability between genetic variant groups

This plot visualises the between genetic variant group variation data presented in Table 2. Between group eta squared values are plotted on a scale of 0% variance to 100% of the variance. These values represent the proportion of variation in phenotypic outcome predicted by genetic variant group. A value close to 0% would indicate a non-specific model whereby different genotypes lead to similar phenotypic outcomes. As value close to 100% would indicate a highly specific model whereby different genotypes lead to different and discrete phenotypic outcomes. The bars indicate 95% confidence intervals.

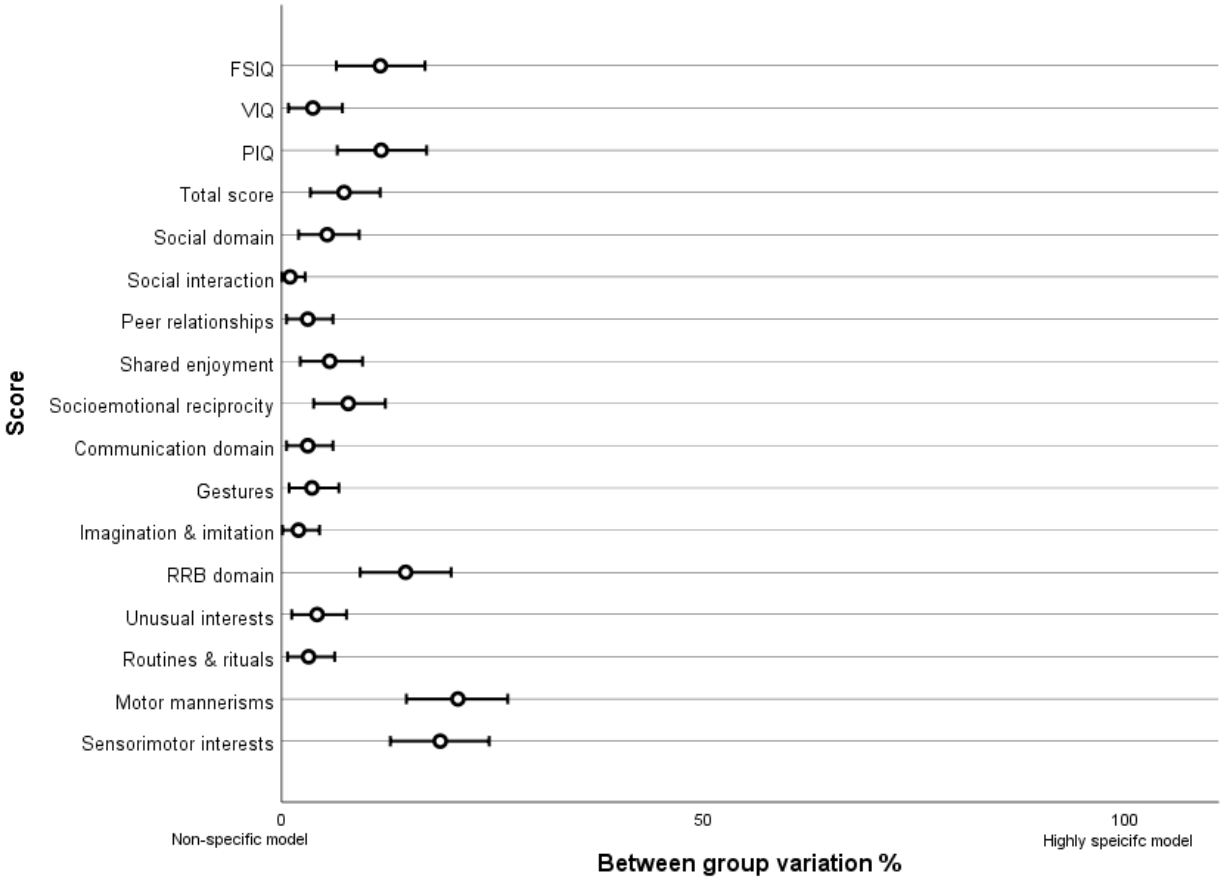


Figure 2: Domain profiles of the genetic variant + autism groups

FSIQ, Full Scale Intelligence Quotient; VIQ, Verbal Intelligence Quotient; PIQ, Performance Intelligence Quotient ; ADI-R, Autism Diagnostic Interview.

2A To visualise how the "genetic variant + autism" groups differed, a heatmap plot was generated by transforming IQ and ADI-R scores of the "genetic variant + autism" groups to z-scores, dendrograms showing the clustering of CNVs and phenotypes were generated using methods described for 3A. Scores for each "genetic variant + autism" were standardized into z scores relative to each other and were adjusted for sex, age and site. The z scores were constructed so that a negative score always denoted a poorer performance. Black indicates a relative deficit in that neuropsychiatric domain compared to other CNV carriers, yellow represents a relative strength compared to other CNV carriers. Hierarchical clustering, for the purposes of presentation (indicated by the dendrogram), was performed using Ward's method and Euclidian distance.

2B To visualise the profiles of the "genetic variant + autism" groups relative to the heterogeneous autism group phenotypic scores were standardised to z-scores, using the mean and SD of the heterogeneous autism group as reference—i.e., the difference in the individual's score and the mean score for the entire autism heterogeneous group was divided by the SD for the heterogeneous autism group. The z-scores were adjusted for sex, age and site. We constructed these Z scores so that a negative score for a -CNV carrier indicated a worse outcome.

Figure 2A

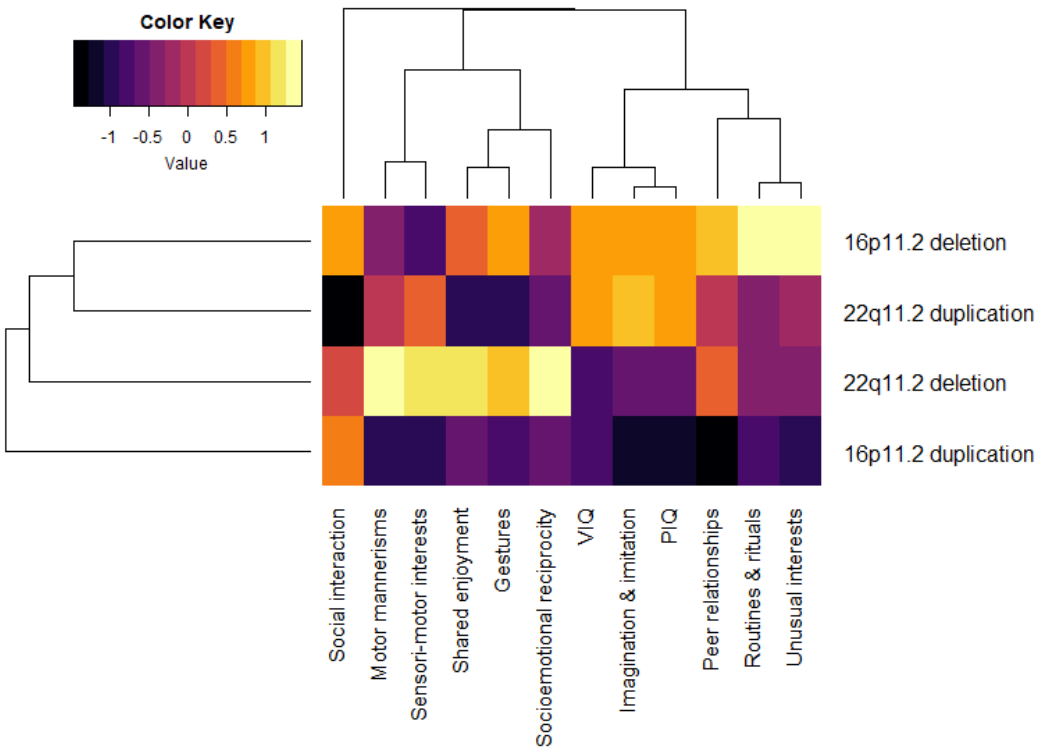
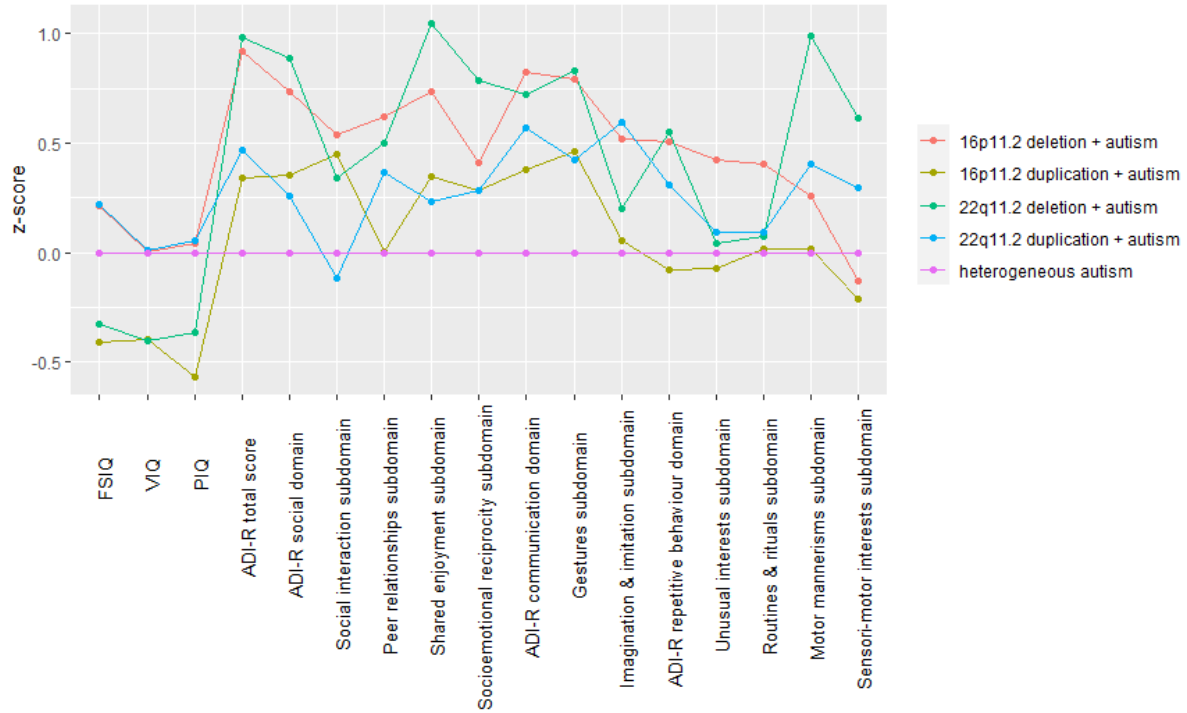


Figure 2B


1.0
0.5
0.0
-0.5
z-score
FSIQ
VIQ
PIQ
ADI-R total score
ADI-R social domain
Social interaction subdomain
Peer relationships subdomain
Shared enjoyment subdomain
Socioemotional reciprocity subdomain
ADI-R communication domain
Gestures subdomain
Imagination & imitation subdomain
ADI-R repetitive behaviour domain
Unusual interests subdomain
Routines & rituals subdomain
Motor mannerisms subdomain
Sensori-motor interests subdomain
16p11.2 deletion + autism
16p11.2 duplication + autism
22q11.2 deletion + autism
22q11.2 duplication + autism
heterogeneous autism