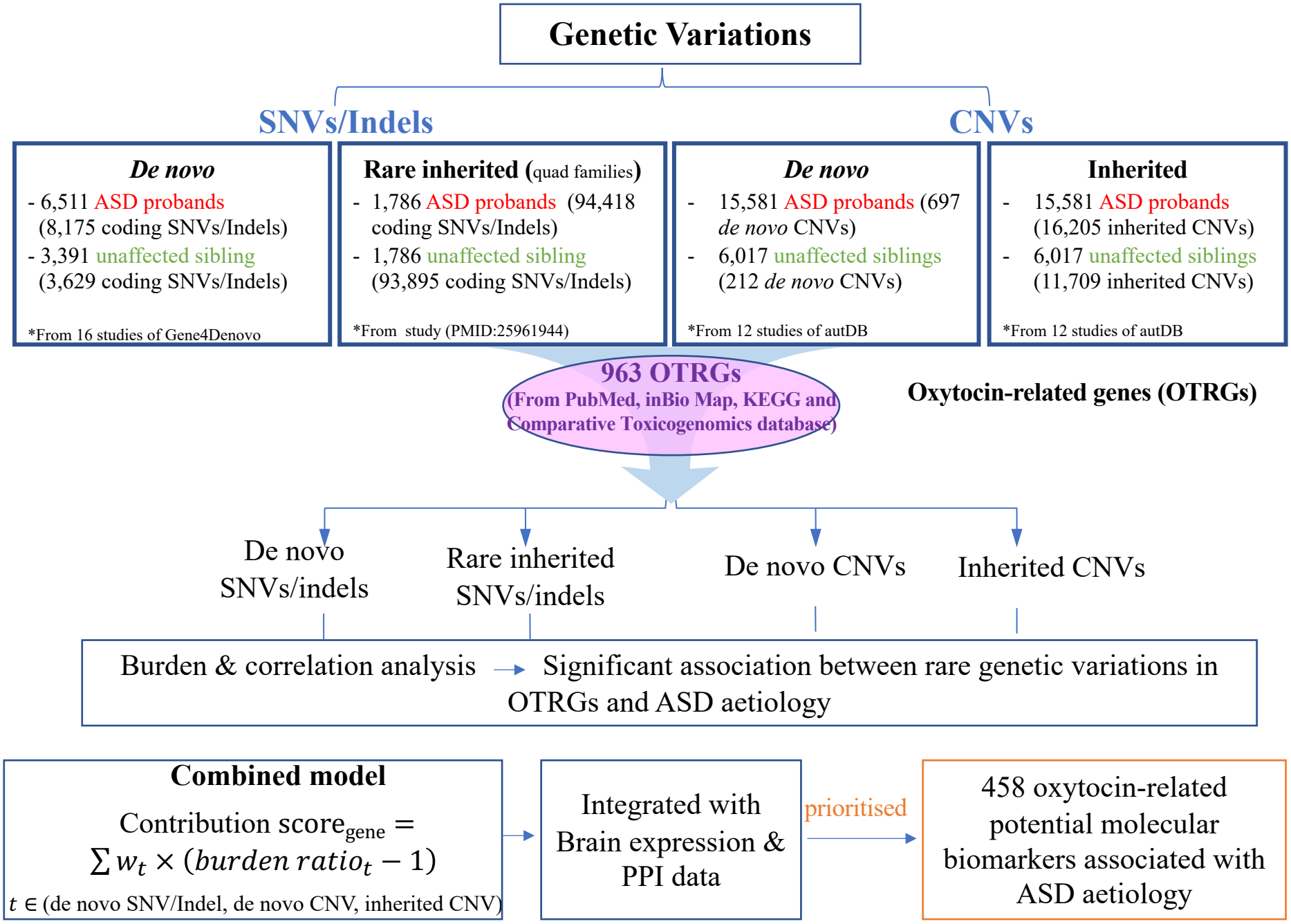
Genetic Variations
SNVs/Indels
CNVs
De novo
- 6,511 ASD probands (8,175 coding SNVs/Indels)
- 3,391 unaffected sibling (3,629 coding SNVs/Indels)
*From 16 studies of Gene4Denovo
Rare inherited (quad families)
- 1,786 ASD probands (94,418 coding SNVs/Indels)
- 1,786 unaffected sibling (93,895 coding SNVs/Indels)
*From study (PMID:25961944)
De novo
- 15,581 ASD probands (697 de novo CNVs)
- 6,017 unaffected siblings (212 de novo CNVs)
*From 12 studies of autDB
Inherited
- 15,581 ASD probands (16,205 inherited CNVs)
- 6,017 unaffected siblings (11,709 inherited CNVs)
*From 12 studies of autDB
963 OTRGs
(From PubMed, inBio Map, KEGG and Comparative Toxicogenomics database)
Oxytocin-related genes (OTRGs)
De novo SNVs/indels
Rare inherited SNVs/indels
De novo CNVs
Inherited CNVs
Burden & correlation analysis → Significant association between rare genetic variations in OTRGs and ASD aetiology
Combined model
Contribution score$_{gene}$ = $\sum w_t \times (burden\ ratio_t - 1)$
$t \in$ (de novo SNV/Indel, de novo CNV, inherited CNV)
Integrated with Brain expression & PPI data
prioritised
458 oxytocin-related potential molecular biomarkers associated with ASD aetiology