

**Combined genome-wide linkage and targeted association analysis of head  
circumference in autism spectrum disorder families**

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**Additional file**

Table 1: Phenotypic characteristics of Utah and AGP samples

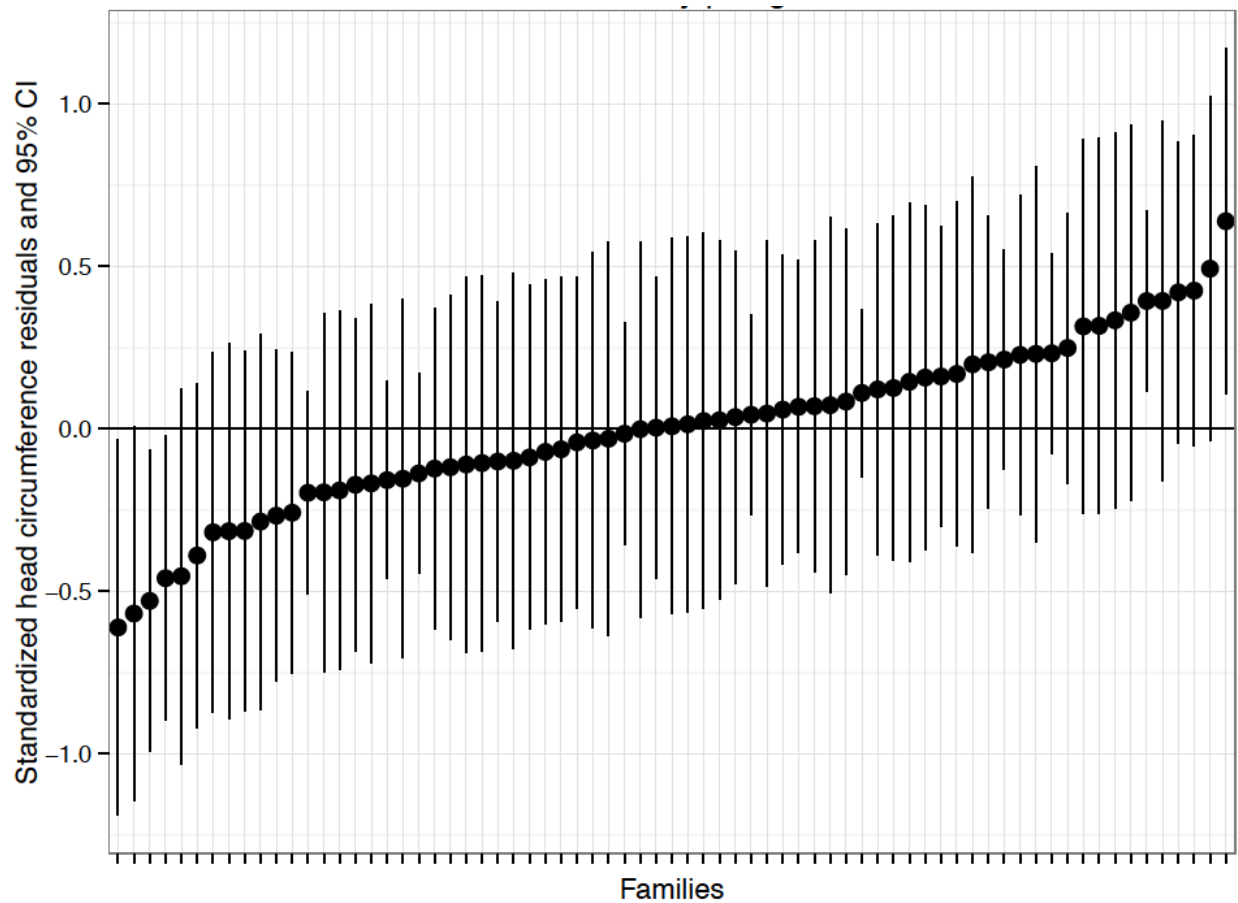
	Utah			AGP		
	ASD (N=198)	Non ASD* (N=469)	All (N=667)	ASD (N=621)	Non ASD* (N=420)	All (N=1041)
IQ Full scale: mean(SD) <sup>1</sup>	85.91 (26.16)	111.99 (12.04)	104.13 (21.22)	87.8 (59.6)	111.2 (14.2)	95.4 (50.4)
IQ Performance: mean(SD) <sup>1</sup>	94.05 (22.22)	113.52 (13.49)	108.13 (18.54)	91.4 (46.6)	110.0 (15.3)	96.6 (41.0)
IQ Verbal: mean(SD) <sup>1</sup>	92.37 (25.21)	108.76 (11.00)	92.34 (17.72)	82.3 (46.1)	109.9 (13.4)	89.9 (41.4)
ADI social: mean (SD) <sup>2</sup>	20.29 (6.73)	NA	NA	NA	NA	NA
ADI verbal: mean (SD) <sup>2</sup>	16.29 (4.23)	NA	NA	NA	NA	NA
ADI non-verbal: mean(SD) <sup>2</sup>	12.47 (2.13)	NA	NA	NA	NA	NA
ADI restricted interests/ repetitive behaviours: mean(SD) <sup>2</sup>	6.49 (2.54)	NA	NA	NA	NA	NA
ADI onset scale: mean(SD) <sup>2</sup>	4.02 (1.16)	NA	NA	NA	NA	NA
ADOS social: mean(SD) <sup>2</sup>	9.10 (3.31)	NA	NA	8.9	NA	NA
ADOS communication: mean(SD) <sup>2</sup>	5.27 (2.29)	NA	NA	5.0	NA	NA
ADOS social +communication total: mean(SD) <sup>2</sup>	14.26 (4.86)	NA	NA			
Vineland Adaptive Behaviour	59.46 (21.46)	NA	NA	55.7 (19.3)	NA	NA

\* Non ASD comprise family members who have generally not undergone clinical evaluation for ASD

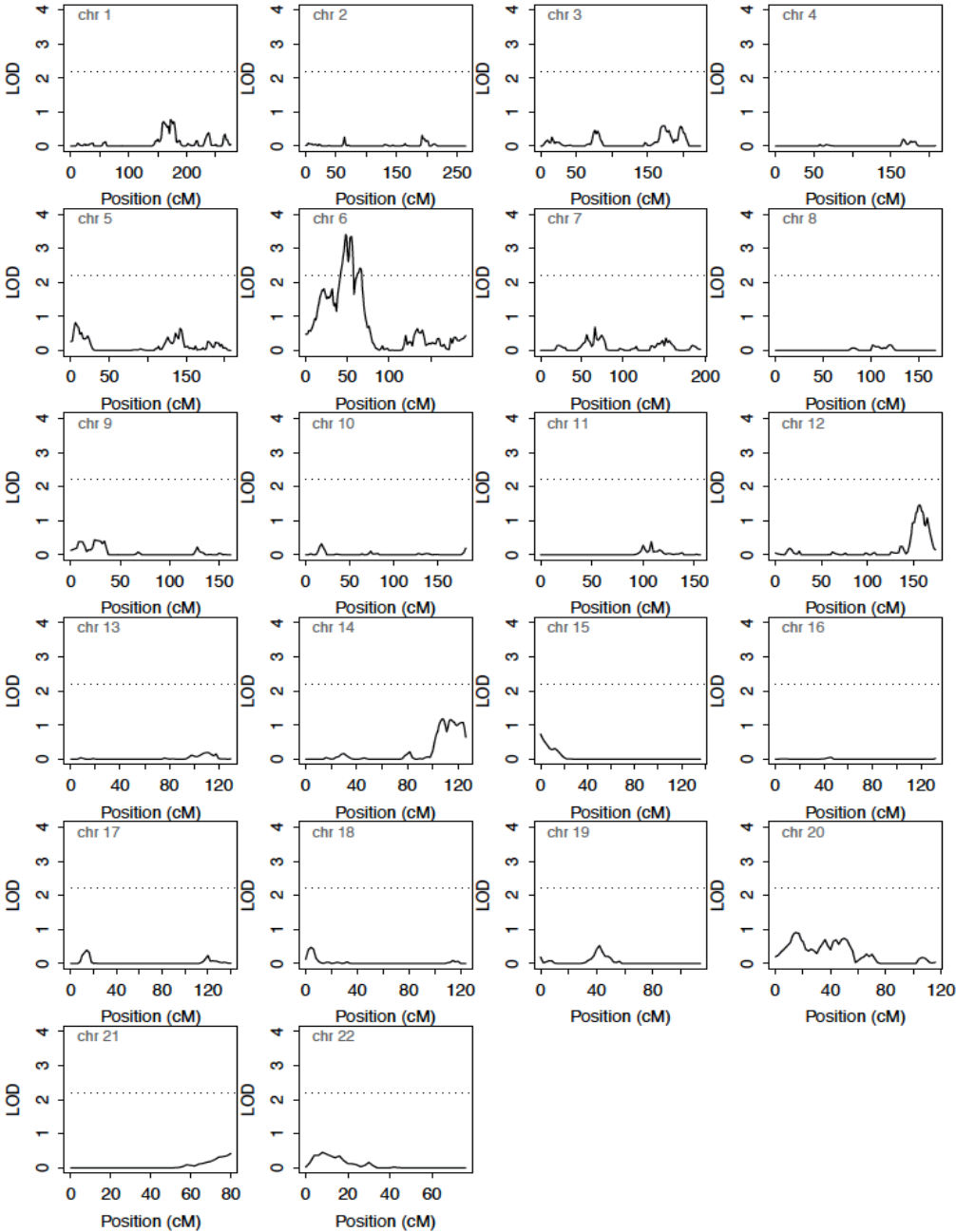
<sup>1</sup> For Utah participants: 177 participants with ASD completed IQ testing. Of these, 147 had testing that included both nonverbal and verbal components; an additional seven participants had only non-verbal IQ testing; 23 had testing that allowed full scale IQ scores only. Of the remaining 21 participants, 19 were not testable and had low adaptive behavior scores and two were not assessed for IQ. 402 non-ASD relatives had IQ testing. Of these, 392 had verbal, non-verbal, and full-scale scores. One non-ASD subject had aphasia, and therefore only had a performance IQ score of 137. The remaining nine non-ASD participants did not have verbal scores due to young age.

<sup>2</sup> 174 participants with ASD had both ADI and ADOS assessments; 21 additional participants had ASD based on ADOS only. The remaining three participants had lifetime clinical diagnosis of ASD supported by Social Responsiveness Scale (SRS) scores in the ASD range.

Supplementary Figure 1: Caterpillar plot of head circumference residuals and 95% confidence intervals by pedigree

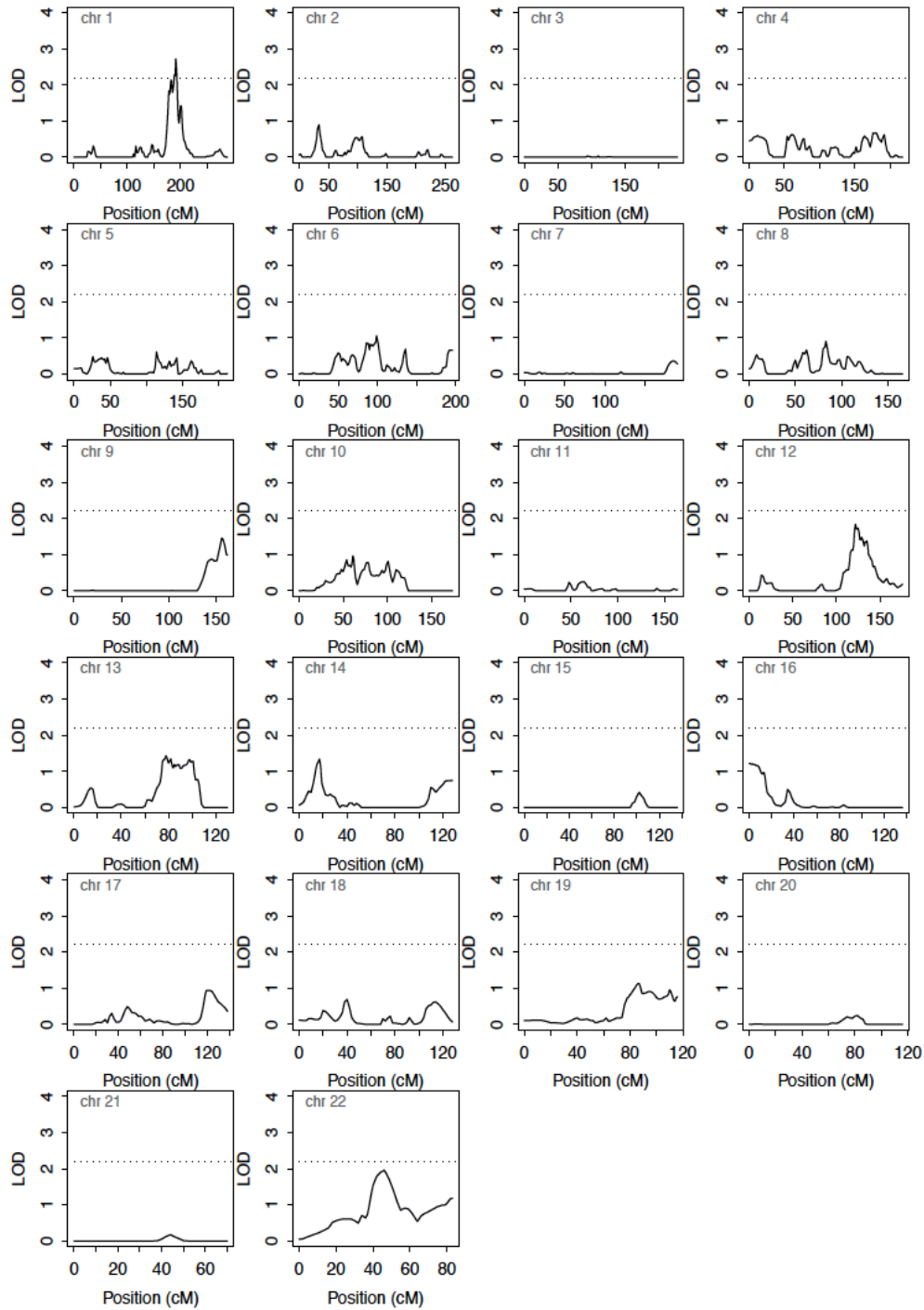


Supplementary Figure 2 Utah pedigrees head circumference genome-wide linkage plots



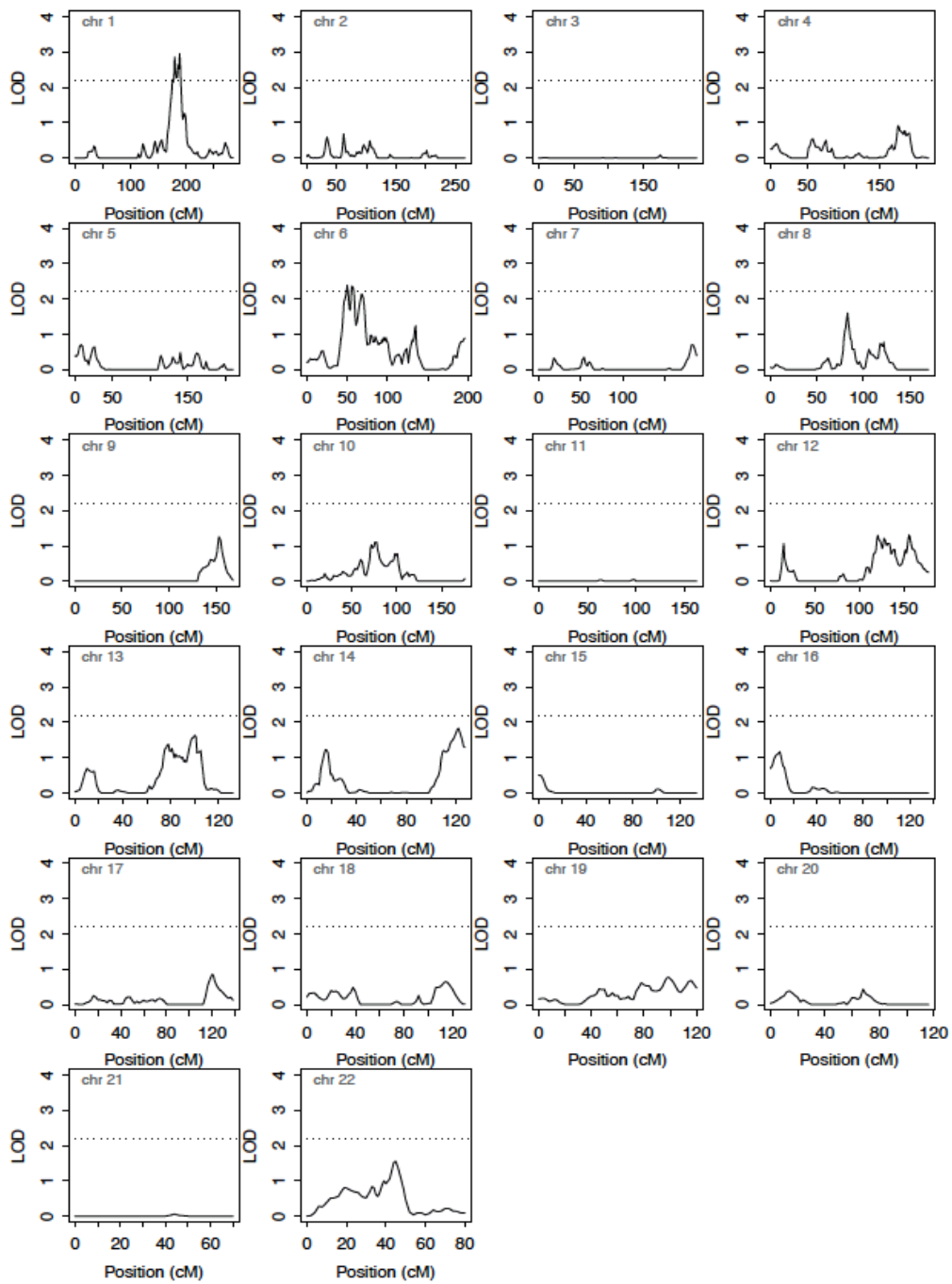
Note: dotted line annotates 'suggestive' linkage according to Lander & Kruglyak (1995)

Supplementary Figure 3 AGP pedigrees head circumference genome-wide linkage plots



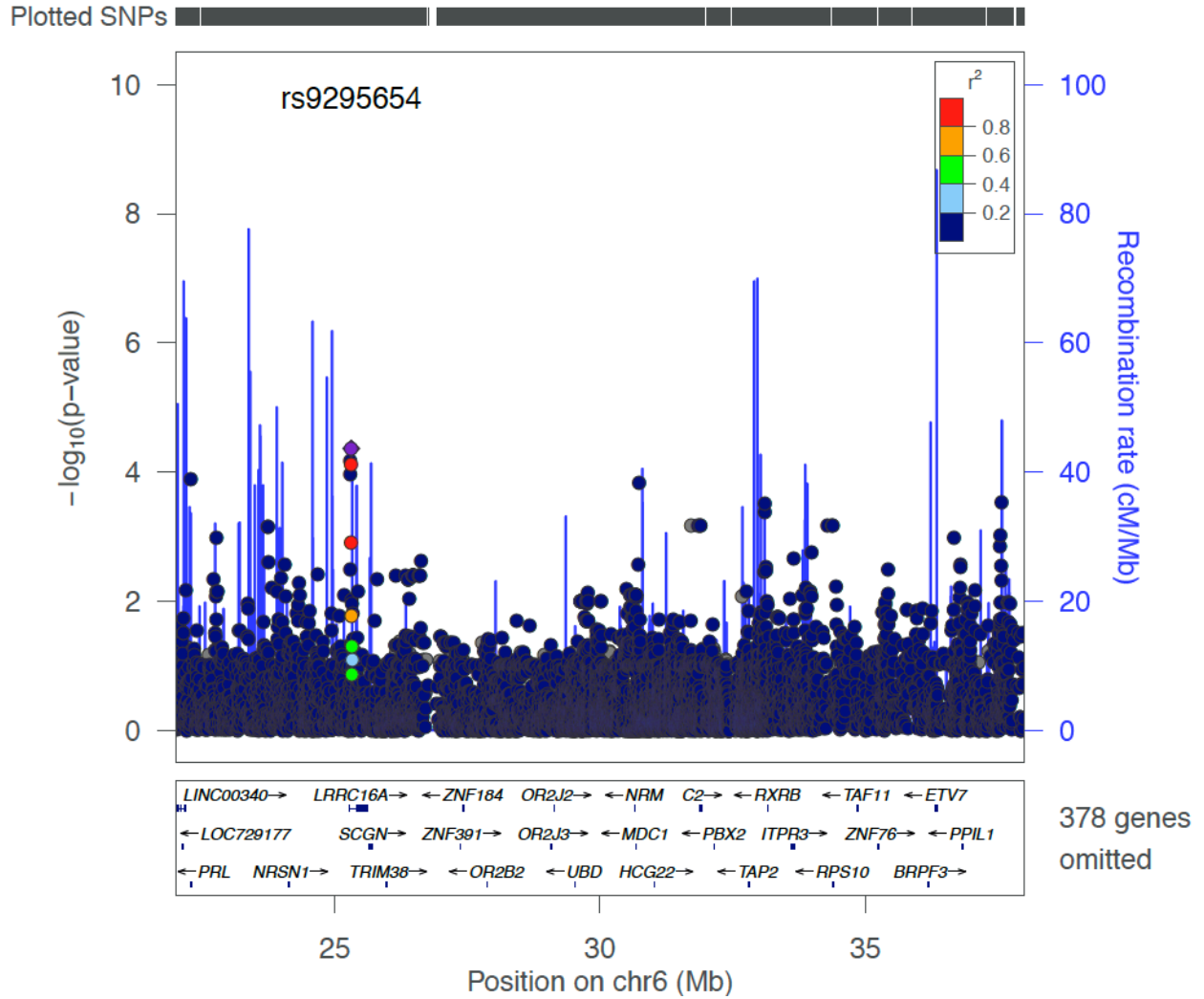
Note: dotted line annotates 'suggestive' linkage according to Lander & Kruglyak (1995)

Supplementary Figure 4 AGP and Utah combined genome-wide linkage plots



Note: dotted line annotates 'suggestive' linkage according to Lander & Kruglyak (1995)

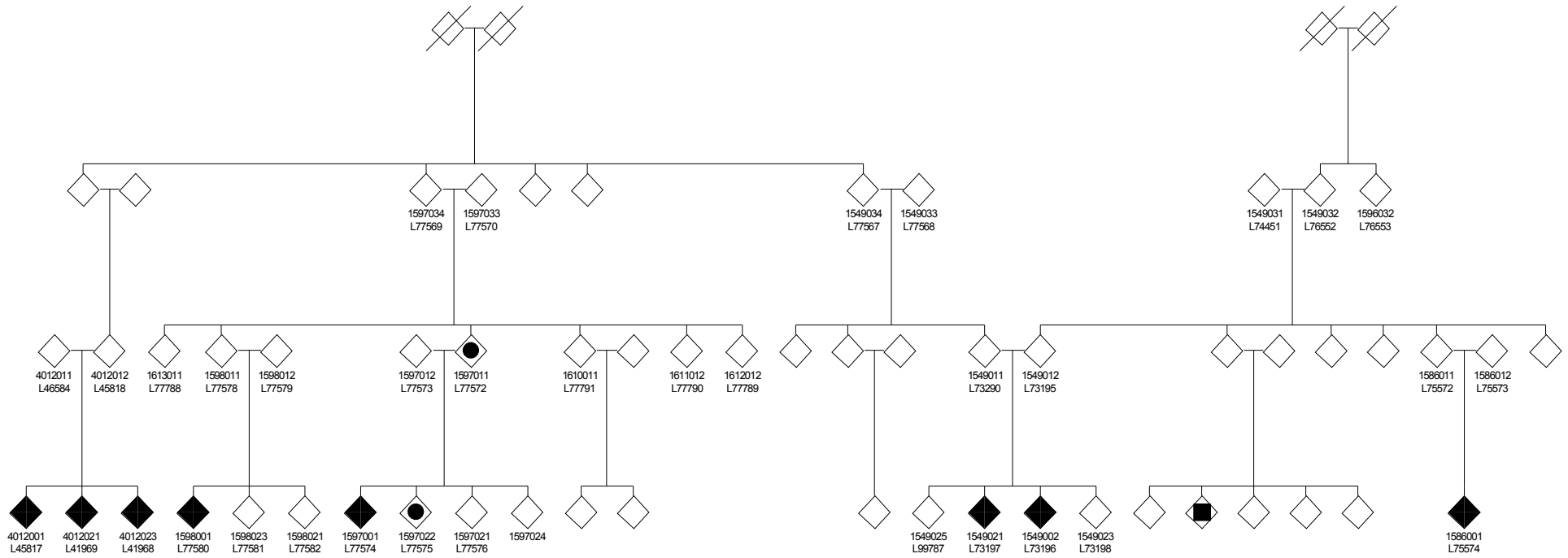
Supplementary Figure 5: Targeted association for complete sample





# Supplementary Figure 6: Family 1

■ Affection Status = Autism    ● Affection Status = PDD    ■ Affection Status = Aspergers



Supplementary Figure 7: Targeted association for Family 1

