Genetic influences on social-cognitive brain structure in childhood: Evidence from Williams syndrome

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INTRODUCTION

- Williams syndrome (WS) is caused by a contiguous deletion of approximately 26 genes on chromosome 7q11.23 [1].
- WS is paired with phenotype characterized by aberrations in social-cognition and hypersociability[2].
- Previous investigations on adults with WS have demonstrated abnormal structural morphology within the insula, orbital frontal cortex and amygdala [3,4].
- Studying children with WS provides insight as to the effect of the WS genetic deletion on the development of the social-cognitive brain [5,6].
- Investigating twins (one with WS and one typically developing) is a compelling method by which to elucidate the effect of genes on social brain development in WS.

METHODS

Participants

- 1 Williams syndrome (WS) twin: female, age 8.32 years
- 1 Typical developing (TD) twin: female, age 8.32 years

Control groups:

- (Behavior) 10 TD 9 f, 1 m, mean age 7.76, SD = 1.51, range = 6.21-10.21
- (VBM) 11 TD, 11 f: mean age 8.28 years, SD = 1.71, range = 6.21 – 10.96
- (FreeSurfer): 10 TD, 6 f, 4m: mean age 8.42 years, SD = 1.87, range = 6.49 – 10.96

Imaging Parameters:

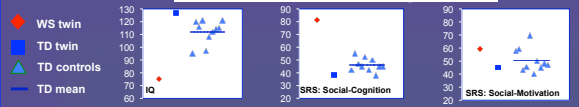
- 3T GE-Signa HDx scanner
- High-resolution T1 SPGR
- TR = 6.4, TE = 2ms
- Flip angle 15°
- FOV = 22cm, matrix = 256x256
- Thickness = 1.5mm

Analysis

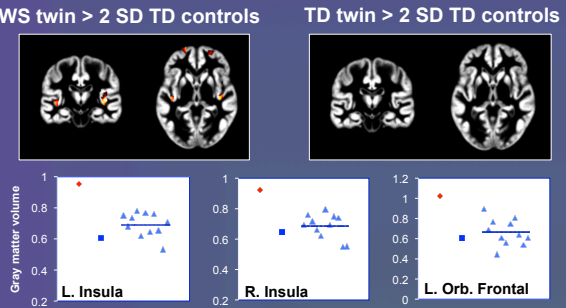
- Behavior: WISC and SRS
- VBM: SPM8
- Freesurfer
- Compare each twin to TD controls
- Significance threshold (>2 SD from mean of controls)
- ROIs: Insula, orbital frontal cortex and amygdala

RESULTS

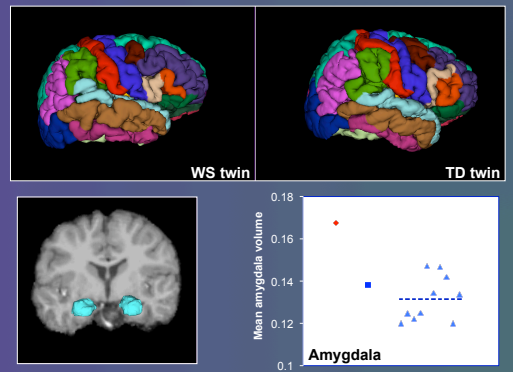
1. Behavior: IQ and Social-Cognition



2. VBM: Insula and Orbital Frontal Cortex



3. Freesurfer: Amygdala volume



CONCLUSIONS

- The WS twin exhibited greater gray matter volume within the bilateral insula and orbital frontal cortex as compared to the TD twin and age matched TD controls.
- The WS twin exhibited greater amygdala volume as compared to the TD twin and age matched TD controls.
- Data provide additional support for a model linking the genetic deletion in WS to aberrations in the neural substrates of social-cognitive functioning in humans.
- Future directions: investigate social brain development in WS using longitudinal approaches.

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