Preliminary results: DS22q11.2 deletion syndrome (DS22q11.2) occurs in 1-2:4000 live births as the result of a 1.5 - 3Mb deletion on chromosome 22. (Murphy & Gammer, 2007).

 Syndrome is characterized by:

- Intelligence scores in "borderline" range (typically PIQ > VIQ)
- Impaired spatial memory, visuospatial attention, numerical and temporal cognition
- Reduced gray and white matter in parietal regions & medial cerebellum consistently reported
- Goal of the present investigation was to assess abnormalities of the amygdala and hippocampus in 7- to 14-year-old children with DS22q11.2 and explore associations with cognitive phenotype.

METHODS

- Data were acquired at 3 scanner sites using a 1.5T Siemens Vision and 2 3T Siemens Trios
  - For each subject, a three-dimensional high-resolution (1mm isotropic) structural MRI was acquired using a T1-weighted MP-RAGE sequence.
  - Although data collection site was not correlated with any of the volumetric measurements it was entered as a covariate in all analyses.

- Total brain volume (gray/white matter and CSF) was calculated using SPM2.

- Neuropsychological guidelines were used to define borders of the amygdala and hippocampus bilaterally and volumes were calculated based on manual tracings of the regions (see Schumann et al., 2004 for details of procedure).

RESULTS - TOTAL BRAIN TISSUE

- DS22q11.2 had significantly less gray and white matter compared to controls.
  - 2 Group x 3 Region MANCOVA w/ site and age entered as covariates, F(1, 68) = 10.12, 8.6, p<.01
  - CSF volume did not differ between the groups (p>.99).

RESULTS - AMYGDALA

- No significant group differences in bilateral volumes of the amygdalae after controlling for differences in gray matter.
  - 2 Group x 2 Hemisphere MANCOVA w/ gray matter, site, and age entered as covariates

RESULTS - HIPPOCAMPUS

- Left hippocampal volume was significantly reduced in children with DS22q11.2, even after controlling for differences in gray matter.
  - 2 Group x 2 Hemisphere MANCOVA w/ gray matter, site, and age entered as covariates

RESULTS - CORTICAL THICKNESS

- DSS2q11.2 had significantly thinner cortical thickness compared to controls.

SUMMARY

- Hippocampal volume was significantly correlated with performance on standardized measures of cognition. All correlations between IQ and hippocampal volume remained statistically significant after entering age, gray matter, and data collection site as covariates.

REFERENCES

- Schumann, C. M., et al. (2004). The amygdala is enlarged in children but not adolescents with autism; the hippocampus is enlarged at all ages. Journal of Neuroscience, 24(9), 6639–6643.

ACKNOWLEDGEMENTS

- Many thanks to the individuals at the Imaging Research Center and the Computational Neuromaging Laboratory; Margie Cabaral, Earl De Guzman, Anna Griffith, Yong He, Victor Laluz, Aaron Lee, Lynndey Marcelino, & Dustin Williams.
- Supported by NIH R01HD46159, R01HD42974, T32-MH073124

Note: Analyses of amygdalae and hippocampi volumes excluded two outliers to ensure that the necessary criteria were met for employing parametric statistics (normality and homogeneity of variance).