Title: Co-occurrence of Autism Spectrum Disorder in Down Syndrome

Authors: Kathryn K. Ostermaier, MD\textsuperscript{1}, Slavica K. Katusic, MD\textsuperscript{2}, Scott M. Myers, MD\textsuperscript{3}, Amy L. Weaver, MS\textsuperscript{2}, Curtis B. Storlie, PhD\textsuperscript{2}, Robert G. Voigt, MD\textsuperscript{1}

Baylor College of Medicine- Texas Children’s Hospital\textsuperscript{1}, Department of Health Sciences Research, Mayo Clinic\textsuperscript{2}, Geisinger Autism & Developmental Medicine Institute, Temple University School of Medicine\textsuperscript{3}

Introduction: Autism Spectrum Disorder (ASD) among children with Down syndrome (DS) is estimated at 1% according to AAP Health Supervision Guidelines. Recent studies report increasing rates of ASD in DS (5 to 48%). However, previous studies are limited by the use of cross sectional cohorts of referred children or by using data from local/ national registries. Our objective was to determine the co-occurrence of ASD among children with DS from a longitudinal, population-based birth cohort.

Method: Karyotype-confirmed DS/ ASD cases were identified from medical and school records from 31,220 subjects in a population-based birth cohort (born 1976-2000) who remained in the community beyond 3 yo and met research criteria for ASD prior to 21 yo. Two research definitions of ASD were explored: (1) Inclusive definition (ASD-R\textsubscript{I}) based on DSM-IV-TR autistic disorder (AD), Asperger’s Disorder, and Pervasive Developmental Disorder (2) Narrow definition (ASD-R\textsubscript{N}) based on DSM-IV-TR AD criteria. Clinical diagnoses of ASD (ASD-C) were also abstracted from medical and school records.

Result: 34 children with DS were identified (1 in 917). 15 (44%) met criteria for ASD-R\textsubscript{N}, 8 (24%) for ASD-R\textsubscript{I}, and 5 (15%) for ASD-C. Of the 5 ASD-C children, 5 (100%) met criteria for ASD-R\textsubscript{I} and 4 (80%) for ASD-R\textsubscript{N}. Of the 15 ASD-R\textsubscript{I} children, (8 (53%) M, 7 (47%) F), 12 had IQ scores available: 7 (58%) had IQ scores ≤ 50, 5 (42%) had IQ = 51-70.

Discussion: Whether considering a clinical, inclusive or narrow research definition, we found ASD rates of 15 to 44% among children with DS from a longitudinal, population-based birth cohort. Our data support vigilant screening for ASD in children with DS.

References/Citations: