

## A Polymorphism of CNTNAP2 Gene is Related to Structural Language Skills in Youth with Autism Spectrum Disorder

Vardan Arutiunian, Ph. D.

Postdoctoral Fellow, Center for Child Health, Behaviour and Development, Seattle Children's Research Institute USA

### Abstract

Although language impairment is not a core symptom of Autism Spectrum Disorder (ASD), it is known that about 75% of children with ASD have a co-occurring language deficit, which may vary from severely impaired language to slightly impaired. At the same time, the biological / genetic basis of these impairments is still not clearly understood. Recent studies have shown that the language profiles in ASD population can be associated with a number of different genetic mechanisms, given the high variability of language skills in these children. One of the candidate genes related to language impairment in children with ASD is the contacting-associated protein-like 2 gene (CNTNAP2), a member of the Neurexin family. Several studies have demonstrated that the polymorphism of CNTNAP2 gene is related to the age of the first word, language comprehension, as well as the severity of autistic symptoms in children with ASD. Additionally, neuroimaging studies (using (f)MRI and EEG/ERP) revealed that the polymorphism of CNTNAP2 gene influences the structural and functional human brain networks which are associated with language and social abilities in general population. However, due to the different assessment tools used in the study it is unknown whether the polymorphism of CNTNAP2 gene is linked to structural language impairment or more general communication abilities. Moreover, the majority of the studies included sex-unbalanced samples and the distribution of genotypes was not equal between males and females; crucially, it has been shown that males and females with ASD can have different profiles with respect to language and communication. The present study aimed to explore the influence of the polymorphism of CNTNAP2 gene on language and communication in a sex-balanced samples of youth with and without ASD. A total of 302 youth aged 7 to 18 years were included in the analysis: 131 youth with ASD (62 females, 69 males), 130 TD youth (64 females, 66 males), and 41 unaffected siblings (US) of youth with ASD (25 females, 16 males). Blood samples were collected to obtain genomic DNA and processed by the Rutgers University Cell and Data Repository (RUCDR) or using standard protocols (Gentra Puregene Blood DNA extraction kit; Qiagen); language abilities were measured with the Clinical Evaluation of the Language Fundamentals - Fourth Edition, and communication skills were screened with Vineland Communication scale - 2. The results showed that the polymorphism of CNTNAP2 was related to language but not more general communication abilities of youth with ASD.