

Abstract

Autism spectrum disorders (ASD) are heterogeneous group of neurodevelopmental disabilities characterized by antisociality and atypical behavioral patterns. Its etiology is very complex, autism is usually formed by combining many factors. One of the causes may be genetic (gene mutation). It is known about 450 candidate genes for ASD so far. Minority of these genes occur in loci which are affected by cryptic rearrangements. These rearrangements significantly contribute to manifestation of this disorder. Pathologies they cause, lead to syndromes with high penetrance for ASD such as Angelman/Prader-Willi or DiGeorge syndrome. Other loci are found on chromosome 1, 2 or 16. Due to short time of studies of cryptic rearrangements, phenotypic variability and number of patients we can expect more researches in the future. These researches are expected not to overlook the impact of the aberrations on formation of autism spectrum disorders.