Abstract

Schizophrenia is a severe mental disorder with high heritability and complex genetics which interacts with environmental factors and leads to a wide range of symptoms. The emergence of modern cytogenetic and molecular genetic techniques has allowed uncovering one of the possible causes - cryptic chromosomal rearrangements. The size of rearrangements, also known as microdeletions and microduplications, is under 3–5 Mb. Aberrations may affect multiple genes and their gene dosage. The research of cryptic rearrangements in association with schizophrenia began in 2008 with the identification of three pathogenic aberrations. Over time studies have identified more cryptic rearrangements and new studies supporting or not supporting their role in the disorder have been published. Research of the candidate genes and their possible interactions has also been conducted. It is hypothesized that schizophrenia is caused by pathologically changed brain connectivity, in which the changed gene dosage by cryptic rearrangements may play a role. The research is in its beginnings, and we can expect the identification of new rearrangements. Further research may lead to a better understanding of the origin and symptoms of schizophrenia, and play a role in prenatal diagnostics and treatment.

Key words: cryptic rearrangements, schizophrenia, microdeletion, microduplication, candidate genes, copy-number variant