







Title: Torn networks - common pathways of neurodevelopmental

disorders

Author: Dorota Hoffman-Zacharska

Department of Medical Genetics; Institute of Mother and Child,

Warsaw, Poland

Abstract:

Neurodevelopmental disorder (NDD) is an umbrella term, that groups a wide range of diseases. Clinically, this term defines in psychiatry disorders with onset in developmental period and characterized by developmental deficits in field of personal, social academic and occupational functions, such as intellectual disability (ID), developmental delay (DD), autism spectrum disorders (ASD) etc. However, considering the etiological criterion most of them involve the impaired neural development and neuronal network formation. This broader form of definition includes spectrum of epilepsy/epilepsy syndromes, as well as schizophrenia, disease of brain development with clinical consequences, which may remain latent form many years.

NDDs are predominantly genetic in origin and are classified into two groups – large group of individually rare genetic disorders and group of idiopathic cases of currently unknown cause.

A well-functioning brain is characterized by good spatial organization and a functionally well-balanced neural network. Recently, the introduction of high-throughput techniques such as NGS and the use of Trios analysis led to the identification of quite a large group of rare NDDs' genes. As it turns out, in the case of NDDs genes are involved in signal transmission (ion channels, synaptic proteins), cellular and nuclear metabolism, as well as encode structural proteins and are related to the division of neurons and their migration. The expression of these genes is subject to spatial and temporal regulation both in the pre- and early postnatal period, building a functional network. Now, by development of sophisticated cellular model systems that recapitulate early human neurodevelopment with spatiotemporal resolution like in organoid cultures, we will be able to better understand these processes.