Article: 1866
Topic: 53 - Mental Retardation

NOVEL CANDIDATE GENOMIC LOCI FOR MENTAL RETARDATION

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Introduction: Genetic disorders underlie a significant number of cases of mental retardation (MR). However, the traditionally used routine karyotyping is not able to detect small but often clinically relevant aberrations. High-resolution genome-wide microarray technologies may help to increase the detectability of MR etiology.
Objectives: To improve MR diagnostics.
Aims: To identify the novel CNV regions and susceptibility genes in MR patients.
Methods: Large chromosomal rearrangements, biochemical defects, and known monogenic syndromes accompanied by MR were ruled out for 71 patients. Further the genome-wide analysis using CGH Microarray Kits 4×44K and 8×60K (Agilent Technologies, USA) was performed.
Results: Eight novel CNVs in 9 patients not described in healthy individuals and not associated with any disease were indentified implicating regions at 11q22.3, 5q33.1, 3p26.3, 15q11.1-q11.2, 15q22.2, 1q25.1-q25.2, 7q21.3, 2q12.3. They include candidate genes *DDX10* (putative RNA helicase), *IL17B* (cytokine, primarily localized to neuronal cell bodies), *CNTN6* (may play a role in the formation of axon connections), *TLN2* (plays a role in cell adhesion and recycling of synaptic vesicles), *TNR* (an extracellular matrix protein, expressed primarily in the central nervous system), *ASTN1* (a neuronal adhesion molecule), *PON1* (detoxifies organophosphate neurotoxicants), and *SLC5A7* (choline transporter in the brain and periphery).
Conclusions: Previously unknown CNVs, containing genes responsible for proper central nervous system functioning, were detected in 13% of patients. The novel CNVs and genes identified by aCGH may help discovering new etiological mechanisms of MR. This study was supported by European Community's Seventh Framework Programme, CHERISH (project 223692) and by Federal Program (grant 8727).

https://doi.org/10.1016/S0924-9338(13)76826-3 Published online by Cambridge University Press