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3P26.3 – CANDIDATE LOCI OF INTELLECTUAL DISABILITY

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Introduction: To date 56 reciprocal microdeletion/microduplication syndromes have been described. Due to intensive application of microarray technologies new submicroscopic rearrangements are being published. The reciprocal rearrangements are particularly valuable, since they allow to determine the dosage-sensitive pathogenic genes.

Objectives: To improve ID diagnostics.

Aims: To identify novel candidate loci of ID.

Methods: We performed the genome-wide analysis for 79 patients with idiopathic ID using CGH Microarray Kits 4×44K and 8×60K (Agilent Technologies, USA). Pathogenically significant cases were confirmed by qPCR.

Results: We present two patients with microdeletion (369 kb) and microduplication (766 kb) at 3p26.3 containing the only gene - *CNTN6.* The microduplication was inherited from apparently healthy father. The child with microdeletion was an orphan. Recently, the microduplication in 3p26.3 (containing *CNTN6*) has been shown to be associated with autism spectrum disorders (ASDs). Contactin 6 is also suggested to play a neuroprotective role in ischemic injury and contribute to granule cell maturation and/or synaptic formation in the developing cerebellum. Considering the experiments with mice we found myotonic syndrome, late development of sit and walk ability in the anamnesis, current fine motor skills impairment and dysarthria in patient with dup3p26.3. His IQ is 47. Dysarthria was also observed in the patient with del3p26.3 (IQ 55).

Conclusions: Obviously, *CNTN6* can be a novel pathogenic gene associated with ASDs, ID, and motor functions impairment. This study was supported by EU Seventh Framework Program, CHERISH project no. 223692 and by Federal Program of Ministry of Education and Science of Russian Federation no. 8727.

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