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Dissecting the Heterogeneity of Autism: Focus on Phelan-McDermid Syndrome

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Introduction: Autism spectrum disorders (ASD) are a group of neurodevelopmental disorders that show delays and deficits in the development of multiple brain functions, which are characterized by social communication, poor language development, and restricted and stereotyped patterns of interests and behaviours. ASD affects about 1-2 % of the population and are considered to be highly genetic in nature. Structural variations of chromosomes have been identified in some ASD individuals, most common on chromosome 7q, 15q and 22q.

Objectives: 1-To present a systematic literature review of the natural history of individuals with 22q13.3 deletion syndrome, Phelan-McDermid syndrome (PMS). PMS, increase awareness of different phenotypes 2- Correlation of clinical manifestations of PMS with hypothesized underlying biological mechanisms 3-Rational for novel treatments is inferred through translational neuroscience approaches.

Methods: We have conducted a systematic literature review of the natural history of individuals with PMS, including both cross-sectional and long-term longitudinal analyses and correlation with hypothesized underlying biological mechanisms, including roles in regulation synaptic development, function, and plasticity. This systematic review includes the basis for a promising common pathway for ASD pathogenesis and the clinical implications of novel therapeutic strategies inferred through translational neuroscience approaches.

Results: This systematic review, therefore, outlines the: (1) Pathophysiological basis and clinical manifestations of PMS; (2) PMS pre-clinical models and applications to ASD; and (3) clinical implications of novel therapeutic strategies.

Conclusions: A promising common pathway for ASD pathogenesis and rational for novel treatments is inferred through translational neuroscience approaches. Neurobiological basis for lithium treatment is indeed supported by experimental results and current clinical findings.

Disclosure: No significant relationships.

Keywords: Autism; Phelan-McDermid Syndrome; Phelan-McDermid Syndrome; autism

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Clinical and psychological features of the brain organization of mental activity in children with autistic disorders

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Introduction: Autism spectrum disorders (ASD) – nosological continuum of genetically and clinically heterogeneous mental disorders united by complex violation of mental development, social interaction and behavior.

Objectives: Identify the features of brain functioning in children with ASD.

Methods: Neuropsychological diagnostics (Zh. Glozman), results acoustic brainstem evoked potentials (ABEP), mathematical methods. The study involved 48 children with diagnoses (ICD-10): F84. 0, F84. 1, F84.5, aged 3 to 8 years (average age 4.18 years).

Results: Neuropsychological examination revealed the main Indices of the functioning of brain blocks: the Index of activation and energy components of activity (I Index), the Index of the right-hemisphere holistic strategy of information processing (Index II-rights), the Index of the left-hemisphere analytical strategy of information processing (Index II-left), the Index of programming, regulation and control of activity (III Index). Then the children were divided into groups ($p < 0.001$): 1 group – 10 children with high I Index ($W = -6.03$); 2 – 20 children with high Index II-rights ($W = -5.74$); 3 – 18 children with high III Index ($W = -2.32$). Correlation analysis showed: for group 1 difficulties in the perception of auditory information are characteristic; for group 2 – reduced level of control over the course of mental activity, difficulties in automating thinking and speech, coordination of movements; for group 3 – relationship is not manifested.

Conclusions: Phonemic perception causes the greatest difficulties in children of group 1. Children of group 2 are characterized by difficulties in the formation of motor speech skills, a decrease in voluntary activity and control over it.

Disclosure: No significant relationships.

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I am the best and I have everything in life

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Introduction: Megalomaniac ideas in a patient with limited intellectual functioning may be due to the psychotic clinic or be the result of their disability.