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Problems in Diagnosing Autism Spectrum Disorders in the Irkutsk Region

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Abstract

The purpose of this study was to evaluate the effectiveness of methods for studying the neuropsychiatric development of children in regards to screening for the diagnosis of autism spectrum disorders (ASD) with the example of educational institutions in the Irkutsk region.

Methods and Results: Two groups of children in educational institutions of the Irkutsk region regional center were studied: 187 children of preschool age (from 5 to 6 years) and 154 children studying at school (from 7 to 11 years). This study used the methods of neuropsychiatric research used by the pediatric service and the methods of screening diagnosis of autism spectrum disorders (ASD). It was shown that the level of obvious anxiety of schoolchildren (according to the scale developed by A.M. Prihozhan) was higher than in preschoolers and was 10.3±0.1% in girls and 10.6±0.2% in boys. Bad appetite was observed in 50.3±3.7% of preschool children and in 56.5±4.0% of schoolchildren. The selective appetite was observed in 16.6±2.7% of preschool children and in 7.1±2.1% of schoolchildren. The study of hyperactive traits showed that only 5.3±1.6% of parents of preschool children noted hyperactivity in their children while educators considered that 22.5±3.1% were hyperactive. The parents noted hyperactivity in 9.7±2.4% of children, and the teachers noted hyperactivity in 21.7±3.4% of children. The intellectual development of children, according to the Raven test, showed that the average IQ was observed in 47.6±3.7% preschoolers, and below-average IQ in 52.9±3.7% preschoolers. At the same time in schoolchildren, below-average IQ was found in 48.7±4.0% and average IQ in 51.3±4.0%.

Conclusion: The study of the development of children's mental processes and behavior is not informative for the early detection of ASD. Likewise, the screening methods for the detection of ASD known today and available to the pediatric service and based on the detection of social and communication disorders are poorly informative. However, if we consider ASD as a manifestation of genetic and cytogenetic pathology, we should look for screening methods in the field of genetics.(**International Journal of Biomedicine. 2021;11(3):337-341.**)

Key Words: autism spectrum disorders • neuropsychiatric development • screening methods

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Introduction

The diagnosis of autism spectrum disorders (ASD) has been discussed more and more frequently in scientific and public circles as of late.(1) Unfortunately, despite the almost century-old history of autism, our knowledge of it does not allow us to provide high-quality medical and pedagogical assistance, nor does it allow us to diagnose ASD in time. Attention should also be paid to the problem of conflicting opinions among different categories of specialists regarding the prevalence of ASD in children. Because of this, psychiatrists rarely rush to make a diagnosis until the clinical picture becomes obvious for its formulation, missing the time span for treatment and rehabilitation. At the same time, the parent community is divided into two camps: those who still hope that the signs of neuropsychiatric development disorders in the child will disappear over time, and are in no hurry to seek psychiatric help, and those whose children were diagnosed after many years of observation.(2)

According to the data of European researchers, between 5 to 11.8 children with ASD per 10,000 children

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are registered.[3] In the United States, there are registered up to 60 per 10,000 children.[3,4] With this in mind, the opinion of Russian researchers on the prevalence of ASD is very different.[5]

Due to the regulatory documents adopted in Russia, pediatric care almost completely skips diagnosis of ASD. The clinical guidelines for ASD published in 2020 recommend that diagnostics be carried out almost exclusively on the basis of behavior analysis and psychological tests.[6]

An early diagnosis is necessary in the first 18-24 months of life for the successful correction and treatment of ASD.[7] However, the diagnosis of ASD in Russia occurs much later, almost when the full clinical picture of the disease unfolds—when the child has already entered school and difficulties in adapting to school education are revealed, or even as late as when puberty starts.

As has been noted above, at present, psychiatrists in Russia do not use actual screening methods for ASD when examining a child for the first time at 2 years of age. In our study, we attempted to analyze the state of diagnosis of ASD in the Irkutsk region and to determine the suitability of the selected methods for diagnosing the neuropsychiatric development of children for screening studies of ASD.

The purpose of this study was to evaluate the effectiveness of methods for studying the neuropsychiatric development of children in regards to screening for the diagnosis of ASD with the example of educational institutions in the Irkutsk region.

Methods

Two groups of children in educational institutions of the Irkutsk region regional center were studied: 187 children of preschool age (from 5 to 6 years) and 154 children studying at school (from 7 to 11 years). The new clinical guidelines for the diagnosis and treatment of ASD published in 2020 [6] recognize three groups of disorders as key manifestations of ASD: disorders in the field of social interaction; communication (anomalies in communication); and limited, stereotypical, repetitive behavior.

In addition to these specific diagnostic features, children with ASD often have a number of non-specific problems, such as phobias, sleep and eating disorders, aggression, and autoaggression, which should be defined as comorbid conditions for ASD. Therefore, the study examined and evaluated the indicators of children's behavior, the characteristics of their personality and behavior, the presence of aggressiveness and hyperactivity (by interviewing parents and teachers using the evaluation scale for hyperactivity),[8,9] characteristics of sleep and nutrition, intellectual development of children (with the help of progressive Raven matrices),[10-13] as well as mental performance.[8]

The neuropsychiatric development of children was studied to identify the morphofunctional abnormalities. The assessment was carried out according to the protocols of the pediatric service. The motor sphere was studied, assessing reflexes and the accuracy of movement coordination; the sensitive sphere, determining pain, temperature, tactile, proprioceptive sensitivity, and stereognosis; the vegetative sphere, assessing skin sweating, temperature, trophism, and dermography, and the orthoclinostatic test.[12,13]

Summarizing the data of the assessment of neuropsychiatric development, groups of children were identified in accordance with the indicators, using the method of distribution of neuropsychiatric development groups adopted in Russia:

Group 1 - Neuropsychiatric development corresponds to or outstrips age

Group 2 - Initial abnormalities in neuropsychiatric development

Group 3 - Pronounced deviations

Group 4 - Severe deviations

The study was conducted by observing children as well as by interviewing children and parents.

Personal anxiety was studied using the scale of explicit anxiety according to A. M. Prihozhan.[11] Screening diagnostics of ASD used the CARS scale (Childhood Autism Rating Scale, translation and adaptation by Morozova and Dovbnya).[14]

The study was conducted in accordance with ethical principles of the WMA Declaration of Helsinki (1964, ed. 2013). Written informed consent was obtained from the participant's parent/guardian.

Statistical analysis was performed using the Statistica 10 software package (Stat-Soft Inc., USA). Analysis of the distribution of values obtained was performed using the Kolmogorov-Smirnov test. The mean (M) and standard error of the mean (SEM) were deduced. For data with normal distribution, inter-group comparisons were performed using Student's t-test. Differences of continuous variables departing from the normal distribution were tested by the Mann-Whitney U-test. The frequencies of categorical variables were compared using Pearson's chi-squared test. A probability value of $P<0.05$ was considered statistically significant.

Results

When studying anxiety in preschoolers, it was found that children on the scale of obvious anxiety had 9.6±0.3 points, in boys slightly more – 9.8±0.3 points. The study of anxiety in schoolchildren showed that the level of obvious anxiety is higher in them than in preschoolers and was 10.3±0.1 points in girls and 10.6±0.2 points in boys.

The interpersonal anxiety was 4.6±0.4 points in preschool children and 5.4±0.2 points in schoolchildren, self-assessment - 3.2±0.2 points in preschool children and 4.9±0.4 points in schoolchildren ($P<0.05$). The interpersonal anxiety was higher in schoolgirls than in boys (4.2±0.6 vs. 2.8±0.4 points, respectively).

The use of the method of projective diagnostics of anxiety according to A.M. Prihozhan (using situational images), supplemented by sociometric studies, revealed a direct relationship between anxiety and low self-esteem in schoolchildren (r=0.57).

The observations during wakefulness and daytime sleep showed that 63.6±3.5% of children in kindergarten were constantly active during wakefulness, 20.3±2.9% were passive and 16±2.7% were irritable. Among schoolchildren,

the indicators of the nature of wakefulness were distributed as follows: active (49.4±4.0%), passive (23.4±3.4%), irritability during wakefulness (27.3±3.6%).

The distribution of individual character traits in preschool children was as follows: disinhibition (19.8±2.9%), cruelty (24.1±3.1%), aggressiveness (36.9±3.5%), and shyness (10.7±2.3%). About 35.8±3.5% of children were easily trained, 9.6±2.2% were not trained, 23.5±3.1% of children showed curiosity. Other individual traits were distributed as follows: affectionate (28.9±3.3%), kind (32.6±3.4%), rude (25.7±3.2%), contact (27.8±3.3%), obsessive (21.9±3.0%), and fearful (15.0±2.6%) (Fig. 1).

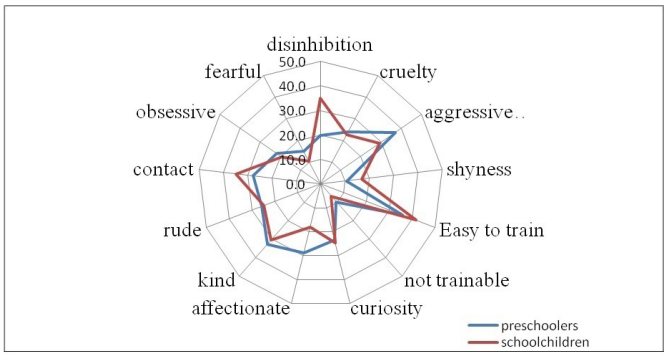

Fig. 1. *The distribution of individual character traits in children*

Among schoolchildren, the individual traits were distributed as follows: lack of inhibition (35.1±3.8%), cruelty (22.7±3.4%), aggressiveness (29.2±3.7%), shyness (16.9±3.0%). Easily trained children consisted of 41.6±4.0%, not trained - 6.5±2.0%, and curiosity was shown by 24.7±3.5% of children. Other individual traits were distributed as follows: affectionate (18.2±3.1%), kind (30.5±3.7%), rude (24.7±3.5%), contact (31.8±3.8%), obsessive (19.5±3.2%), and fearful (10.4±2.5%) (Fig. 1).

In view of the fact that sleep disorders can be observed in ASD, studies were conducted on the nature of sleep and ability to fall asleep in children. Due to the peculiarities of the work of educational institutions, sleep was studied only in preschool. It was shown that about 33.2±3.4% of children fell asleep poorly and 81.3±2.9% had shallow sleep.

According to the available literature, the appetite in children with ASD is often unstable or selective. We also studied the appetite in children to clarify the nature of eating disorders in our research. The following results were obtained in preschool children: good appetite (7.0±1.9%), bad appetite (50.3±3.7%), unstable appetite (9.6±2.2%), increased appetite (15.5±2.6%), and selective appetite (16.6±2.7%) (Fig.2).
In schoolchildren, the appetite traits were distributed as follows: good appetite (7.8±2.2%), bad appetite (56.5±4.0%), unstable appetite (14.9±2.9%), selective appetite (7.1±2.1%). The selectivity of appetite was mainly manifested by the avoidance of meat and dairy products.

The identification of indicators of attention deficit using the hyperactivity rating scale showed that only 5.3±1.6% of parents noted hyperactivity in preschoolers while caregivers considered that 22.5±3.1% children were hyperactive. Among schoolchildren, the parents noted hyperactivity in 9.7±2.4% children, the teachers noted hyperactivity in 21.7±3.4% of children.

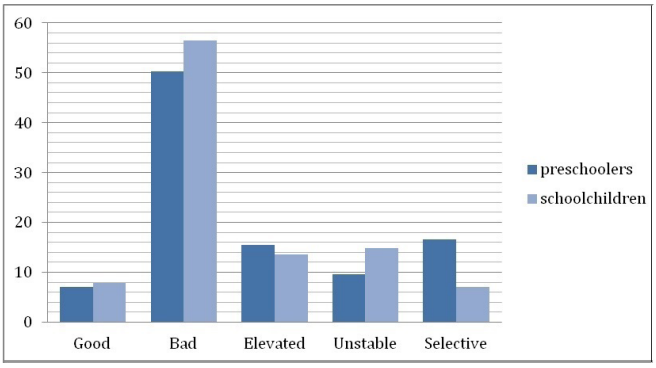

Fig. 2. *The indicators of appetite in children*

The lagging neuropsychiatric development seen in ASD may be one of the first signs of the development of the disease. The obtained data allowed us to divide the children into groups of neuropsychiatric development (Table 1).

Table 1.
Neuropsychiatric development groups of children

Neuropsychiatric development group	Preschool children	Schoolchildren
Group 1	69.5±3.4	55.2±4.0
Group 2	28.3±3.3	40.3±4.0
Group 3	2.1±1.1	3.9±1.6
Group 4	-	0.6±0.6

The study of the levels of intelligence in preschoolers by the Raven test showed that average IQ was found in 47.6±3.7%, below-average IQ in 52.9±3.7% of cases (Figure 3). In schoolchildren, the results were as follows: below-average IQ in 48.7%±4.0 of cases and average IQ in 51.3±4.0% of cases. It should be noted that a low level of intellectual development was not detected, which is a very important result in our study. High levels of intellectual development due to age characteristics were also not identified.

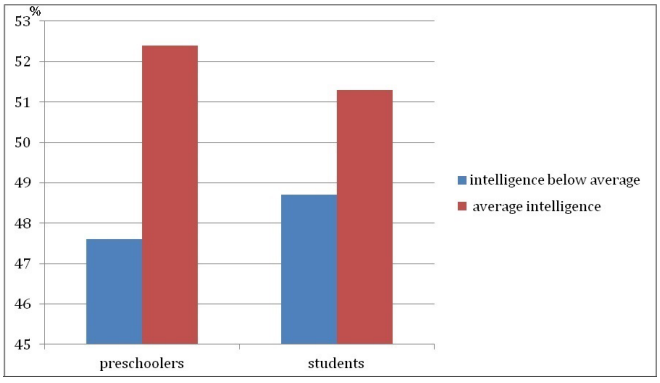

Fig. 3. *The levels of intelligence in children (the Raven method)*

When analyzing the results of the quality of the completed tasks of the Raven test, it was found that children on average perform 38.7±1.2% of the tasks. The indicators of the levels of attentiveness, imagination, and visual difference (discrimination), as well as the establishment of relationships between the figures, was quite high (6.7±1.1 points). Best of all, the children of both groups revealed an analogy between the figures – 7.3±1.1 points. The level of dynamic mindfulness and the establishment of dynamic analogies were slightly lower and amounted to 5.2±0.9 points. In schoolchildren and preschoolers, we found the lowest level of the ability to perceive quantitative and qualitative changes and arrange them according to the regularity of the changes (2.8±0.6 points), the ability to observe the complex quantitative and qualitative development of kinetic, dynamic series, showing abstraction and dynamic synthesis (1.1±0.3 points).

The screening diagnostics of ASD[13] showed that all the examined children scored less than 30 points, i.e. no manifestations of autistic disorders were found. However, 5.3±1.6% of the examined kindergarten children scored between 20 and 25 points, which may indicate that there are individual violations. Deviations from the norm were observed according to such criteria as attitude to people, adaptation to changes, nervousness and fears, verbal and nonverbal communication, level of activity, and consistency of intellectual response.

There were also no obvious signs of autistic disorders among schoolchildren. In the group of schoolchildren, deviations from the norm were more manifested by nervousness and fears, a violation of verbal communication, and the level of activity.

Contrary to expectations, there were more adverse signs in girls than in boys, but the severity of the sign was higher in boys, although the small sample size does not allow us to judge the statistical significance of the differences.

The study of the mental performance of preschool children revealed that the productivity index was 5.4±0.5, errors - 2.4±0.7, and the number of lines viewed - 11.6±0.4. In school children, the productivity index was 11.7±0.6, errors - 1.6±0.6, and lines viewed - 13.2±0.3. The girls looked at slightly fewer lines than the boys, but they also made fewer mistakes.

Discussion

The diagnostic criteria for ASD (defined by ICD-10 and required for diagnosis) are grouped as follows: qualitative disorders of social interaction and changes in communication, limited, repetitive stereotypical patterns in behavior and interests (activities), non-specific problems (fears, phobias, agitation, sleep disorders and eating habits, rage attacks, aggression, and self-harm). One of the diagnostic indicators is the manifestation of these conditions before the child is three years old.[12,13,15]

In our study, in addition to the accepted screening methods, other methods were used that allow us to identify disorders of the neuropsychiatric development of children. However, despite the methods used, it was not possible to identify children with signs of autistic disorders. As noted earlier, foreign and domestic authors claim that, currently, there are between 5 to 11.8 children with ASD per 10,000 children in European countries,[3] and up to 60 per 10,000 children in the United States.[3,4] According to the data provided by the Psychological, Medical and Pedagogical Commission (PMPC) operating in the Irkutsk Region, 66 children with a confirmed diagnosis of ASD are enrolled in educational institutions in the Irkutsk region (as of September 1, 2020), which is 1.14 per 10,000 children.

The epidemiological data on the incidences of autism among adults was not available until recently in Russia. The first results, according to Rosstat, were obtained at the end of 2015: 14,692 children (under the age of 18), which is 0.06% of all children of this age in Russia, and 102 adult patients with ASD, which is less than 0.001% of the entire adult population in Russia; in 2016: 18,224 patients under the age of 18(0.08%) and 96 (<0.001%) adults. In recent years, thanks to pediatric observations and the dissemination of information about the problem of autism, the diagnosis of ASD in children in Russia occurs much earlier than in the past, increasing the number of detected cases of the disease.[16]

Our research has shown that the available assessment tests (as screening tests) and methods for studying the neuropsychiatric development of children are too painstaking and uninformative.

Most children with neuropsychiatric disorders, despite the presence or absence of an established diagnosis, attend specialized or private educational institutions. In educational institutions in general, it is almost impossible to identify children with mild ASD using existing methods. In view of this circumstance, the statistical data differ greatly both in different countries and in the territory of the Irkutsk region in comparison with the data on Russia as a whole.

Another problem in the diagnosis of autism spectrum disorders is the age of children at which it is still possible to make a diagnosis of ASD. For successful treatment, early diagnosis is necessary (preferably earlier than 18 months), and diagnostic criteria, according to the new clinical recommendations,[6] are violations in the field of social interaction; communication (communication abnormalities); limited, stereotypical, repetitive behavior that cannot be formed at the age of less than 18 months. This is a clear contradiction in the formulation of the diagnosis. Our study showed that social and communication skills in preschool and primary school children are not sufficiently formed due to age and may hide pathological violations of these qualities.

Another aspect of the problem of diagnosing autism spectrum disorders is the lack of understanding of the pathogenesis of ASD.[5,17,18] But if we proceed with the postulate that autistic disorders are just a syndrome of impaired social and communication skills, which occurs from certain genetic and epigenetic abnormalities, then the diagnostic criteria for ASD can be genetic disorders. Now, a number of genetic diseases characterized by autistic disorders are already known. These include Fragile X syndrome, also called Martin-Bell syndrome, ,tuberous sclerosis (mutations in either the TSC1 gene or the TSC2 gene), Rett syndrome (mutations in the MECP2 gene),

phenylketonuria (mutations in the PAH gene), Down syndrome (trisomy 21), Prader-Willi syndrome (a deficiency of paternal gene expression on chromosome 15q), Angelman syndrome (a deficiency of maternal gene expression on chromosome 15q), Smith-Magenis syndrome (deletion of chromosome 17p11. 2), DiGeorgi syndrome (22q11.2 deletion syndrome), Phelan-McDermid syndrome (22q13.3 deletion syndrome), Kleefstra syndrome (9q34.3 microdeletion syndrome). Other genetic abnormalities characterized by autistic disorders are likely to be identified. In this aspect, research on somatic health is very important,[19] as well as the study of the pathogenesis of autistic disorders.[19,20]

If we continue to assume that ASD is a manifestation of genetic abnormalities, then screening and diagnosis using currently known methods that lie in the plane of psychiatry and neurology, most likely will not lead to success.

Conclusion

New methods, which identify risk groups for the development of neuropsychiatric disorders at the early stages of a child's development and timely development of optimal algorithms for specialized psychoprophylactic care for each child, could present a promising future in screening diagnostics of ASD in children. This is especially important for the conditions informing education and upbringing when the load on the psyche is increasing many times. In the Irkutsk region, the problem of screening children with ASD remains unresolved—the number of detected cases of ASD most likely does not reflect the actual situation.

In our opinion, it is worth reviewing the known diagnostic approaches to ASD, perhaps changing the routing of patients, transferring diagnostic preferences from the field of psychiatry to the field of genetics. In accordance with this, treatment methods may be found in the field of gene therapy.

The increase in the number of children with ASD cannot but cause concern in scientific and public circles. The search for diagnostic criteria for autistic disorders, despite numerous studies in this area, including our research, remains an urgent area of scientific research.

Competing Interests

The authors declare that they have no competing interests.

References

1. Malinina EV, Sergeev VA. The actual issues of psychiatry, organic mental disorders and epilepsy: the materials of the scientific and practical conference. Chelyabinsk: Publishing House «Pirs»; 2012. [In Russian].
2. Maltinskaya NA. [The history of the development of the autism doctrine]. Scientific and methodological e-journal "Concept." 2017;1:11. [Article in Russian].
3. Duchan E, Patel DR. Epidemiology of autism spectrum disorders. Pediatr Clin North Am. 2012 Feb;59(1):27-43, ix-x. doi: 10.1016/j.pcl.2011.10.003.
4. Levy SE, Mandell DS, Schultz RT. Autism. Lancet. 2009; 374(9701):1627-38. doi: 10.1016/S0140-6736(09)61376-3.
5. Dolgikh VV, Starovoitova TE, Pogodina AV, Mikhnovich VI, Rychkova LV. [Somatic health of children with autism]. Pediatriya - Zhurnal im G.N. Speranskogo. 2014;93 (5):150-153. [Article in Russian].
6. Clinical Guidelines for Autism Spectrum Disorders (approved by the Ministry of Health of the Russian Federation). 2020. [In Russian].
7. Fesenko YuA, Shigashov DYu. [Early childhood autism: a medical and social problem]. Health - The Basis of Human Potential: Problems and Ways of their Solution. 2013;(1):271-273. [Article in Russian].
8. Kuchma VR. The hygiene of children and adolescents: Textbook. Moscow: GEOTAR-Media; 2008. [In Russian].
9. Kuchma VR, Tkachuk EA, Tarmaeva IY. [Psychophysiological state of children in conditions of informatization of their life activity and intensification of education]. Gig Sanit. 2016;95(12):1183-8. [Article in Russian].
10. Prikhozhan AM. The anxiety in children and adolescents: psychology of anxiety and age dynamics. M: Moscow; 2000. [In Russian].
11. Mazurin AN. Propaedeutics of children's diseases: textbook, the 3rd ed. St. Petersburg: Foliant; 2009. [In Russian].
12. Istratova ON. Psychodiagnostics: a collection of the best tests. The 7th ed. Rostov: "Phoenix"; 2010. [In Russian].
13. Schopler E, Reichler RJ, DeVellis RF, Daly K. Toward objective classification of childhood autism: Childhood Autism Rating Scale (CARS). J Autism Dev Disord. 1980 Mar;10(1):91-103. doi: 10.1007/BF02408436.
14. Childhood Autism Rating Scale, CARS (Schopler E, Reichler RJ, DeVellis RF, Daly K).Translation and adaptation by Morozova TYu, Dovbnya SV. Moscow; 2011. [In Russian].
15. Dyusova SG, Kaibalieva GH, Shevtsova OS. Features of mental development in early childhood autism: specific disorders of perception, speech, thinking, fears. Collection of scientific works "In the world of scientific discoveries." (September 28, Taganrog; 2015). 2015;1:28-31. [In Russian].
16. Skvortsov IA. Children's autism in neurology. Moscow: MEDpress-inform. 2020. [In Russian].
17. Nason B. [Core Challenges of Autism. Sensory aspects of autism]. Autizm i narušenie razvitiâ = Autism and Developmental Disorders. 2016;14(3):42-48. doi:10.17759/autdd.2016140304. [Article in Russian].
18. Albitskaya ZhV. [Early childhood autism - problems and difficulties of primary diagnosis in interdisciplinary interaction]. Medical Almanac. 2016; 2 (42). [Article in Russian].
19. Darenskaya M, Rychkova LV, Gavrilova OA, Kolesnikova L. Signs of oxidative stress parameters in boys of different ethnic groups with exogenous-constitutional obesity. Free Radical Biology and Medicine. 2018;128:47-60. doi: 10.1016/J.FREERADBIOMED.2018.10.082
20. Tkachuk EA. Martynovich NN, Rychkova LV, Polyakov VM. [Problems of diagnosis of autism spectrum disorders in children]. Pacific Medical Journal. 2020;4 (82):35-43. [Article in Russian].