Screening, identification and evaluation of autism spectrum disorders in primary health care

Praćenje, identifikacija i evaluacija spektra autističkih poremećaja u primarnoj zdravstvenoj zaštiti

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Introduction

Autism spectrum disorders (ASD) is a terminology present in the literature over the past two decades. It represents a group of disorders with common basic expressions in different degrees which are pervasive developmental disorders such as children's autism, Asperger syndrome and pervasive disorder – not otherwise specified.

The reasons for separating and assembling these three disorders are that they have common basic behavioral manifestations (clinical signs) and are relatively well defined as autistic syndrome, although the degree of expression may vary in different extent. Precisely, these differences in the presence and degree of basic clinical characteristics represent the spectrum of autism disorders.

Conceptualization of disorders

The terminology “autism spectrum disorders” clearly reflects the current perception and understanding of autistic disorders and is a new concept accepted in this area.

This concept is based on a research which results showed that children's autism, Asperger syndrome and autistic disorder not otherwise specified, are just different kinds of a basic, autistic, behavioral phenotype.

Basically, these are clinical signs which correspond to the clinical picture of children's autism, the one first described by Kaner in 1943, when he revealed eleven children in a group of mentally retarded children who showed “a strange and extreme isolation distance”.

It also includes a disorder that the Austrian psychiatrist Hans Asperger discovered in 1944, describing amongst children's autism, children whose cognitive abilities and verbal skills were at a much higher level. This entity got its name – Asperger syndrome after him.

Terms of conceptualization of disorders, which means, comprehension and understanding of the disorder, have evolved over years in accordance with the degree of knowledge and understanding of the disorder, such as the prevailing doctrines.

For a long time, children's autism was placed in a group of psychotic disorders in childhood. Children's autism, as a separate clinical entity, showed up for the first time in 1980, when the National Council of the American Association for autism suggested a third edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-III classification) to promote a rather different conceptual approach. Children's autism has been since then (and is now) understood and considered not as a psychosis but as a disease with damaged flow of normal developmental processes in the social, cognitive and psychological spheres. In other words, children's autism is a pervasive developmental disorder.

All “borderline” cases, in terms of incomplete clinical signs or associated with other disorders (comorbidity) since 1987 (in DSM III-R) are classified as not otherwise specified autistic disorder.

Asperger syndrome, as a separate entity, is given the “right of citizenship” only in 1994.

Recognizing the presence of basic common “woof” in these three disorders makes the conceptualization of these disorders as a unity, defining them as autism spectrum disorders (ASD).

In both major international classifications, International classification of diseases and disorders (ICD-10) and DSM-IV-R, these disorders are still today in the group of pervasive developmental disorders (with Rett syndrome and disintegrative...
Epidemiological data for this group of disorders suggest that the prevalence is about 6 in 1000. It must be said that the data, at different times and from research to research, varied depending on the “width” and “strictness” of diagnostic criteria and some paramedical factors (involvement in special education, permanent housing ...). Research of disorders in relation to sex showed the ratio varying from 2:1 to 6.5:1 in favor of boys. This ratio is even more pronounced in cases where Asperger syndrome is found in only one girl towards 15 boys.

Autism spectrum disorder is a behavioral phenotype defined by clinical signs. That is why it is diagnosed as a behavioral disorder 1–5.

It should be emphasized that this grouping of ASD includes extremely heterogeneous phenotypes, including also those with vaguely defined expression as well as the prominence of different levels of behavioral manifestation, especially in the middle of the spectrum.

The essence of behavioral disturbances is expressed by inability to make reciprocal relations and communication with other human beings in a way that it is normal or common. The core feature of the disorder consists of qualitative impairment of social interaction, qualitative impairment in communication and stereotyped, restrictive and repetitive forms of behavior, interests and activities.

Qualitative impairment in reciprocal socio-emotional reactions

These children lack the awareness of the presence of or feelings towards others, do not notice or treat them as pieces of furniture; passively uses people to meet their needs, has no interest in other children, do not participate in playing with others or make other children participate as mechanical “extensions”; do not register other persons’ feelings (sadness, joy); show the lack of emotional responses to the messages of other people. These children do not possess or, if they do, their ability to imitate is damaged (they do not know to wave as a greeting, they mechanically imitate others’ actions out of context).

Qualitative impairment in verbal and non-verbal communication

There may be a lack of communication in any way (mime, gesture, spoken language) and/or markedly abnormal nonverbal communication, without using gaze “eye to eye”, without facial expression, body position and gestures to initiate or modulate social interaction. If speech is present, there are clear abnormalities in the production of speech (including volume, height, rhythm, intonation; for instance: squeaking or questioning melody etc.); in form and content (there are a lot of stereotypes and repetitive uses of speech – echolalia and metalalia; speech in the third person; there is idiosynkratic use of words and phrases); inability to start or to continue a conversation with others despite the preserved speech.

Extremely narrow repertoire of activities and interests and repetitive and stereotyped forms of behavior

These children have very limited interests, such as the interest for important dates, driving schedule, telephone directory. They insist on maintaining the routine in detail (they have to go the same way to the kindergarten or a shop) and show extreme anxiety at a minimum change in the environment (change of lamps or flower pots, for instance). The lack of imagination when playing is evident. The game is stereotypical, often repetitive, and even bizarre. There is fascination (permanent preoccupation) in touching parts of objects or unusual objects (they can play for hours with the lid of some pot or a screw, or can touch the texture of some material). Motoric stereotypes are characteristic: bizarre stereotypical hand movements in the form of knocking, clapping or flapping, or swinging, bouncing and rolling the whole body.

These are the elements of clinical and diagnostic criteria for children’s autism. All of these symptoms must be present, but the degree of their prominence varies from case to case, and this is what causes variation in the expression of the disorder.

In addition to these specific diagnostic criteria, autistic children have a range of non-specific problems from fear, sleep and eating disorders, temper tantrums and aggressiveness, to especially self-injury.

Certainly the most important “extra” coexisting conditions, which does not make the core, are the cognitive deficit (general developmental slowing down). Intellectual abilities of children with autism vary in a wide range from normal ones (measured non-verbal techniques) to those whose intellectual development was heavily disturbed. The question of cognitive deficits, especially of mental retardation, was actualized in the 90s of the last century. In fact, attitudes that over 90% of children with this disorder show delayed mental development are somewhat questioned: the results of recent studies have shown that this percentage is under 50 and that it is actually about the difficult assessment of cognitive abilities of these children (assessment instruments and professional training are questioned). It must not be forgotten that people with Asperger syndrome are, by definition, characterized by normal intelligence. As some children with heavily impaired intellectual development can show “strange islands of skills on their general cognitive level, it is considered that cognitive abilities of children with autism do not follow the usual course of development.

In one third of children suffering from autism, epilepsy occurs during childhood or adolescence. Neurological and somatic tests in these children are normal.

Diagnosis

Diagnosis is based on the fact that the clinical manifestations (behavioral manifestations) fulfill the diagnostic criteria which are given in two of the world’s leading classifications: ICD-10 and DSM-IV R 4,5.
Diagnostic criteria (according to DSM-IVR) for children with autism–pervasive developmental disorder, are 5.

A. Qualitative impairment of social interaction; Qualitative impairment in communication; Restricted repetitive and stereotyped forms of behavior, interests and activities.

B. Delayed or abnormal functioning in each area, starting before 3 years of age: social interaction, use of language for the purpose of communication and symbolic or imaginative playing.

C. Disorder that does not fit in Rett syndrome and children’s desintegrative disorder.

Diagnostic criteria (according to DSM-IVR) 5 for Asperger’s syndrome are: Qualitative impairment of social interaction; Restricted repetitive and stereotyped forms of behavior, interests and activities; Disorder causes clinically significant impairment in social, occupational and other important areas of functioning; No clinically significant slowdown in the development of language; No clinically significant lags in cognitive development or the development of appropriate self-help skills, adaptive behavior and curiosity about the school environment; The criteria can not be applied to another specific pervasive developmental disorder or schizophrenia. Diagnostic criteria for autism spectrum disorders are shown in Table 1.

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<tr>
<th>Types of ASD</th>
<th>Common disorders</th>
<th>Different disorders</th>
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</thead>
<tbody>
<tr>
<td>Children’s autism</td>
<td>Manifests before 3 years</td>
<td>General slowing down or abnormal functioning in each of</td>
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<td></td>
<td>Characteristic abnormal functioning (limited, stereotyped and repetitive)</td>
<td>the above areas</td>
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<tr>
<td>Asperger syndrome</td>
<td>Non-specific problems: phobias, sleep and eating disturbances, temper tantrums,</td>
<td>There is no clinically significant slowdown in the</td>
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<td></td>
<td>aggression and self-injury</td>
<td>development of speech</td>
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### Etiology

Autism spectrum disorders (ASD) etiology is not known, but today we consider that, although the spectrum of autism disorders is the behavioral phenotype defined by its clinical signs, it is biologically conditioned by neurodevelopmental disorder with a high degree of inheritance 7, 11, 12. In fact, it is considered that the ASD neurobiological disorder occurs in genetically predetermined persons as a result of different unknown factors that act as triggers in the first two trimesters during the intrauterine life. These conclusions are based on facts as follows: Biological studies of central nervous system (CNS) in children with ASD (primarily autism in children) resulted in a number of interesting new findings. “Diversity” of pathological findings greatly hinders their integration into a single theoretical concept. However, different aspects of biological disorders combined

in a coherent sequence, where the surveillance, in accordance with different compatible elements, structures one theoretical model encompassing many aspects.

### Genetic basis

Certainly the most significant considerations are those regarding the genetic basis of this disorder 12. Generally designed by researches of genetics within population and focused researches in the field of molecular genetics (targeted cytogenetics, screening the entire genome within families of children with autism spectrum disorders) they have given more than intriguing and promising results.

Research in the field of population’s genetics 10, 11 has shown that the behavioral manifestations of autism are related, more than randomly (1 of 4 “classic” cases of autism – about 10%), to a single gene disorders (neurofibromatosis, tuberous sclerosis, untreated phenylketonuria, Hurler’s syndrome). The nature of this connection is unknown.

The biggest incentive for the study of genetics of autism at the molecular level is the discovery of the relation between X fragile chromosome (Xq 27.3) and autism (1982). The presence of X fragile chromosome (and typical clinical picture), the so-called FRAXA syndrome, was discovered in 14% of all cases of autistic disorders. Although it is ac-
Neurodevelopmental model

In autism caused by developmental damage of CNS there are two different groups of each of the findings that are the basis for the conceptualization of this model.

The first group – clinical observations of similarity between autistic behavior and behavior of adults in the verified and known syndromes (neglect syndromes in the frontal lobe syndrome, Kluer Bucy syndrome, Korzhakov’s psychosis).

The second group – results of neuroanatomical and pathohistological researches.

Pneumoencephalography (in 1970s) and brain computerized tomography (CT) of patients with autism have shown a primary dilatation of the left temporal horn of lateral chambers in about 25% of these patients. Autopsy studies in patients with autism have shown major changes in cellular composition (number and size of neurons) in the nuclei amigdala and hypocampus (structures that are localized in the mesial temporal lobulus), which is a characteristic of immature configuration and de facto a sign of bacwardness in maturuation.

The second structure where the CNS in patients with autism showed significant changes is the cerebellum (the reduction of Purkinje and granular cells, in the vermis and cerebellum hemispheres, with preserved neurons in the lower olivar nucleus which suggests that this loss of cells occurs during prenatal development). Abnormalities of cerebral cortex were also evidenced.

Based on these findings, it is assumed that these regions are dysfunctional due to immaturity, which is the result of distorted migration of cells in the CNS during the first 6 months of fetal life is the result of early dysfunction caused by disorders of organogenesis con causa ignota.

Different functional MRI studies have shown that individuals with ASD use different cognitive strategies and, in many cases, other brain regions for processing specific information. Consequent damage has shown the connection between different cortical regions in the brain of people with ASD. Deficits in empathy, imitation and speech are due to abnormalities in the functioning of mirror neural systems. These functional brain differences promise intriguing connection between neuroanatomical substrate and the characteristic clinical manifestation in patients with ASD.

Immunological theory

The known facts about the participation of immunological processes as a “major player” in ethyopathogenetics of autism are, so far, unconvincing.

Identical (or very similar) clinical sign is a reflection of existence (involvement, participation) of the same anatomical structures of the CNS at a certain point of development (probably during the first months of gestation). Its consequence is the development of certain types of disorders in various domains of psychological functioning (so-called autistic behavior).

Why are these etiological considerations important if we say that the etiology of ASD is actually unknown?

First of all, because of a certain number (10–20%) of disorders which “look like” – have a clinical spectrum of autistic disorders or are associated with ASD and thereby have a clearly defined etiology (eg untreated phenylketonuria, X fragile chromosome syndrome, neurofibromatosis, tuberous sclerosis...). In some cases these disorders are considered in literature as secondary ASD as opposed to the primary ones with the basically unknown etiology. This is very important from the differential diagnostic point of view.

Screening, identification, evaluation of children with ASD in the primary health care

Basic items which model this issue and are present in the previous presentation are: autism spectrum disorder is a behavioral phenotype defined by its clinical signs (this is why it is diagnosed as a behavioral disorder); ASD is by its nature a neurobiological disorder generated by environmental factors on the genetic susceptible basis (most likely during the first two trimesters of intrauterine life); there are no specific biological tests for ASD.

These are also the guidelines for creating an “action plan” for screening and identification.

Developmental counseling in primary health care is conceived for developing a comprehensive monitoring and evaluation of child’s situation during regular visits to a pediatrician (in child’s 9 months, 18 months, 24 months and 30 months of age). This serves particularly to identify and track children ‘at risk’ (personal history, family history). It also helps to identify and evaluate children susceptible of having the ASD.

Screening

The key part for early identification of this disorder is screening – monitoring milestones of development expected for specific age and tracking of signs of development in the areas of speech, social and communication skills as well. These milestones are monitored in the general population of children aged 0 to 3 years within monitoring in developing counseling centers, and especially in the population of children at risk for ASD (children at risk for ASD are those who have a positive family history or risk factors in the former development). As part of a periodic monitoring of pediatrician through visits (9, 18, 24, 30 months of age) all variations in the development of social relationships, communication and speech are registered.

Identification

What are the steps to make if there is a doubt regarding the existence of symptoms of Autism spectrum disorders as well. For a good assessment, good knowledge of milestone development of social skills, communication and speech is indispensable (Table 2).

Signs of social skills deficits may be specific, but are often subtle and hardly recognized by parents. However,
Stage of alarm ie “danger on the way” of development are: lack of appropriate gaze (“eye to eye”); lack of happiness in terms of exchange to-and-fro (here and there) vocal forms between infant and parents; lack of recognition of mother’s (father’s or a permanent caregiver’s) voice; lack of response to the name; disregard for vokalizations; delayed onset of babbling; decrease or absence of prespeech gestures (showing, waving, pointing); lack of expressions such as “oh oh”; “huh”; lack of interest or response to any kind of neutral term (observation, participation) so. “Joint attention” looks at an alternative facility that is scored, and parents, the social interaction of multiple emotional expressions, voices and other gestures, respond to requests “show around” (nose, mouth ...)

Evaluation

After the diagnostic tests are carried out the evaluation of the results is made.

The evaluation is done at two levels and these are: evaluation of the child where there are manifestations of autism spectrum disorders, evaluation of the procedure in terms of “steps” to undertake in the identification-diagnostic procedure.

Basic principles of evaluation are: step by step evaluation; completeness – integration and analysis of the results; monitoring during a period of time.

When the existence of the disorder is confirmed a new interview with the parents must be performed. Parents are made aware of the child’s problem and the child is referred to children’s psychiatrist. The next step in the specialized institution is a general analysis and the individualized evaluation of child’s capacities. According to it, a program is made of specialized sociotherapeutic intervention and treatment (speech therapist, special teacher).

Evaluation of the child where there are manifestations of autism spectrum disorders. The evaluation is performed by a qualified team which includes a child psychiatrist, pediatric neurologist, psychologist, and a speech and language therapist.

After confirmation of the diagnosis, the child is referred to a specialized institution for comprehensive assessment and intervention. The evaluation process involves a multidisciplinary team consisting of a child psychiatrist, pediatric neurologist, psychologist, speech and language therapist, and a social worker. The evaluation process is performed in accordance with the guidelines of the American Academy of Pediatrics.

New detailed case history of the parents regarding: the child’s development; the observed changes in its development; family case history. Surveillance of the child – intellectual level of functioning, specific developmental delays, behavioral assessment “meeting” the criteria for ASD; child’s tests – testing the existence of dysmorphia and neurological deficits; tests in terms of identification of syndromes whose etiology is known and show the picture or are associated with ASD (X fragile chromosome syndrome, tuberous sclerosis, untreated phenylketonuria, Rett syndrome...); audiological check up – AEP; genetic check up (X fragile chromosome, Mec P2); metabolic screening; electroencephalographic (EEG) examination; magnetic resonance (MR) examination of the endocranium – does not make part of a routine diagnostic.

Conclusion

Autism spectrum disorder is a new concept and approach to a group of disorders that include: children’s autism, Asperger syndrome and pervasive disorder otherwise not specified. These disorders make a range of behavioral phenotypes (given the varying degree of prominence and representation of certain elements) which core makes the inability of reciprocal relations and communication with other human beings in a way that is normal/common.

Relatively homogenous (spectrum) behavioral phenotype, etiological heterogeneity, prevalence increase, gravity of disorders and chronicity (long-term – often for a lifetime) impose as an imperative good knowledge of all elements of this disorder with the aim of early identification. Early detection is a prerequisite for early intervention that can significantly alter the course of disorder and improve the quality of life of children with ASD and their families.

The first “meeting point” and the early diagnosis of this disorder is in the hands of the pediatrician in developmental counseling. Monitoring the development of common milestones expected for age and milestones in the development of social skills, communication and speech is a prerequisite, that is, the first step is screening. Registering variations in the development of social relationships, communication and speech is the second step – the identification of potential ASD in children patients. The third step are the preliminary diagnostic tests, a comprehensive evaluation and assessment.

Three steps phase in early diagnosis of ASD in the developmental counseling is a major step in helping children with ASD. Early diagnosis allows appropriate planning and implementation of individually made behavioral and educational interventions, structured support of professional ASD specialists, program support and family assistance.
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