Chapter from the book *Autism Spectrum Disorders: The Role of Genetics in Diagnosis and Treatment*

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1. Introduction

The term Autism Spectrum Disorders (ASD) came into use in 1988 to stress the fact that symptoms of autism may appear with various intensity – from very mild to very severe. The matter gets further complicated due to the fact that the symptoms occur in different combinations. Autistic deficits include not only “autistic triad”, that is disorders of speech, behaviour and social interactions but disturb also cognitive, and motor abilities as well as emotional functioning. Despite a long-term interdisciplinary studies and detailed descriptions of autistic symptoms diagnosis of ASD still causes a number of diagnostic difficulties. The diagnostic procedure has been made easier thanks to the introduction of two systems of classification - International Statistical Classification of Diseases and Related Health Problems (ICD-10)\(^1\) and DSM-IV-TR\(^2\) - but it still remains a long and arduous process. It happens quite often that the final diagnosis is made in the course of therapy. Assessment of children with ASD must takes into account their spontaneous and reactive behaviours as well as information gained during the interview with their parents. As mentioned above, dysfunctions occurring in autistic spectrum vary to a considerable degree in their intensity. Moreover, they are of a dynamic character and are apt to undergo changes as a result of a course of general development, individual experience, social conditions, undertaken therapy, and efficacy of stimulation. All these may hamper the process of evaluation, especially at the early stages of a child development. Hence, autistic children are often made a diagnosis of mental disability, behaviour disorders, hearing problems as well as strange and eccentric behaviours. To complicate the matter further the above disorders frequently coexist with autism. It is imperative, therefore, to be able to discriminate autism and other developmental disorders since the early diagnosis provides basis for creating effective therapeutic and educational programs.

2. Frequency of ASD occurrence

Recent epidemiological studies show a dramatic increase of the number of persons with autistic symptoms. Thus, the frequency of occurrence of ASD was rated at a level of 4 to 10,000

\(^1\) Proposed by World Heath Organization – WHO, 10\(^{th}\) revision, 1992
\(^2\) Diagnostic and Statistical Manual of Mental Disorders developed by American Psychiatric Association - APA, 2000; a new version V is to be published in May 2013
cases in the years of 1980-90, while in the last decade the number of autistic persons is considered to reach the level of 2 to 1000 persons, and 2 to 1000 persons in the case of Asperger syndrome. At the same time, the population of persons with ASD is rated at the level of 1-2 to 100 cases in the U.S.A., the country in which the most advanced epidemiological studies are carried out. In addition, statistical data show that the increase of a frequency rate of ASD is 10 - 17% a year (Newschaffer et. al., 2007). It may be due to a real increase of the occurrence of autism but might as well reflect refinement of assessment techniques as well as a wider scope of diagnostic criteria, such as lowering down the age of assessment, broadening the scope of diagnostic criteria as well as development of diagnostic tools and techniques. In addition, the awareness of the parents of disabled children has also changed lately.

An analysis of statistical data shows also that ASD disorders are more frequent in boys than in girls. It is estimated as 4,3 to 1, while in the case of Asperger syndrome the occurrence of the disorder in boys rises to 8 in comparison to 1 girl (ICD-10, 1997). There are a number of theories trying to explain such a state of affairs. One of frequently cited is the theory of neurotoxic testosterone. It presumes that due to the unequal development of brain hemispheres in the prenatal stage a very high level of testosterone brings about disturbances in the development of the left hemisphere, which results in a very high occurrence of autistic disorders in boys (Geschwind, Galaburda, 1985a, 1985b, 1985c).

The idea was further elaborated by Baron-Cohen and collaborators (2005). They conducted a longitudinal study, which aimed at revealing a possibility of the influence of so called foetal testosterone (FT) upon the development of a child in the prenatal stage of life. The study included pregnant women in whom level of testosterone was measured in amniotic fluid obtained via amniocentesis in order to evaluate the influence of difference in a level of FT upon a subsequent development of the child. The results described also in the following paper (Auyeung et al., 2009) revealed a negative correlation between FT and the development of language and social skills, and positive correlation with specific traits of autism, such as excessive concentration on details, stereotyped movements, and perseverations. The studies on hiper-mascunalization in ASD are carried on, since the role of the influence of testosterone upon the foetal development needs further verification. Yet, an analysis of androgens influence on the appearance of autistic disorders seems to bring promising results (Chakrabarti et al., 2009).

The above presented data raises a question whether a clinical picture of autistic symptoms in girls differs from that observed in boys, and what - if any - is a specificity of autism occurring in girls. Kopp (2010) noted that autistic symptoms in girls are often neglected by professionals despite the reports of anxious parents. It creates the need of presenting a more detailed analysis of autistic disorders observed in girls. All the more that the most available descriptions concentrate upon the characteristics of autism based upon an analysis of boys are of a rather general nature. Therefore, we decided to perform an analysis of autistic symptoms in autistic girls from the perspective of microgenetic and microdevelopment approaches. We were motivated by the fact that those theories make possible the evaluation of developmental potential of autistic persons. In order to make the clinical picture of autism in girls as complete as possible the description of individual cases will be presented in some detail.

3. Autism spectrum disorder in girls

As mentioned above, most studies published so far give general information, which is mostly based upon an analysis of autistic boys. As a rule the sex of examined subjects is not
taken into consideration, especially as ASD is prevailing in boys. Lately, specificity of ASD occurring in girls has been noted, yet most authors still concentrate upon symptoms characteristic of autism in general, such as tendency to routine behaviours, lack of interest in fantasy games, difficulties in social interactions and/or distractibility leading to learning problems (Knopp, 2010). On the other hand, McLennan and collaborators (1993) report that Loveland observed lowering of general IQ in boys (N = 700), while it was not stated in girls (N = 300). At the same time, it was noted that no use is made of the developmental potential of autistic girls, which may result in lower social and communication capacities and greater difficulties in establishing relations with peers in girls (Lord & Schopler, 1985). In addition, girls score worse than boys both in verbal abilities and visual-spatial tests. According to Nichols (2009) it reflects higher level of expectations concerning social and communication skills in girls, and hence more negative evaluation of observed deviations. Inappropriate behaviours of girls are often interpreted as a way to make others to pay attention to them, while in the case of boys such behaviours are believed to reflect their attempts to get a desired object.

The differences between sexes are also believed to be a result of differences in developmental trajectories. Some authors believe that boys exhibit more difficulties at the early stage of their life, while the difficulties in girls increase in the period of adolescence (Nichols, 2009; Nichols et al., 2008). Symptoms of brain damage, however, are more frequent in girls than in boys, which finds its confirmation in EEG records, which show more irregularities in girls. On the other hand, the autistic girls are better in performing games that require using rules and also show weaker tendency to stereotyped movements than boys (Lord et al., 1982; Nichols, 2009).

Lord and collaborators (1985) point out that longitudinal studies revealed that girls with ASD did not establish any friendly relationships during a period of ten years, while several boys did accomplish it. According to these authors it may be due to the fact that girls tend to be more short-tempered and tearful (see also McLennan et al., 1993). Those discrepancies in behavioural and neurological functioning tend to disappear if the autistic girls are offered appropriate stimulants for their development. Yet, the autistic symptoms often appear again as the years go.

Kopp and colleagues (2010) compared the quality of life of 100 girls with ASD and ADHD aged from 3 to 18 years, and it made them believe that those two types of disorders tend to co-occur, since ADHD was stated in 95% of autistic girls. At the same time, a higher level of fear, sleeping problems as well as a higher risk of depression was noted in both ASD and ADHD. Moreover, comparative studies of girls with ASD and ADHD revealed a regression in development in comparison to healthy subjects of the same age. The dysfunctions were observed both in psychological, motor, and social abilities so they included all aspects of behaviour. It needs to be stressed that a positive influence of environmental factors, especially of appropriate education and family conditions, proved to stimulate the development of autistic girls. Hambrook and collaborators (2008) observed that anorexic girls exhibit lack of empathy and of an ability to systemize as well as other autistic traits. It is emphasized that a distorted pattern of information processing characteristic of anorexia shows a significant similarity to the autistic spectrum. Those difficulties may take various forms such as a lack of cognitive flexibility or stereotyped behaviours. A good example of rigid patterns of response noted both in ASD and anorexia provides an inability to shift a plan of action.
4. Microgenetic approach form developmental point of view

A traditional approach to the study of developmental processes concentrates either on a long period of time (longitudinal studies) or upon groups including as big numbers as possible (cross-sectional studies). In recent years many scholars emphasize the usefulness of research taking into account the scores gained by the same group of children evaluated in short time intervals. Such an approach has derived also from the microgenetic theory. In other words, it enables an observer to monitor the specific moments of transformations in thought and behaviour in contradistinction to classic longitudinal studies, which provide only a general pattern of a change of behaviour in examined subject (Levelly et al., 2005). Flynn and co-workers (2006) enumerate three aspects pointing to the usefulness of a microgenetic approach:

1. It makes possible delineation of a whole range of a mechanism underlying a process of changes.
2. Observations are conducted while the factor causing a change is at work and not only before and after it took place.
3. It is possible to control a moment of passing from applying a stimulus and initiating a change.

At the same time, microgenetic approach enables getting answers to the following questions:

1. Is the instruction understood by the child?
2. Does the child use innovative strategies in solving a given problem?
3. Is the child able to discover a new more effective strategy in the course of action?
4. What is the efficiency of actions undertaken by the child while looking for a proper solution?
5. What amount of time does a child need to solve a particular problem?
6. Is the child able to generalize an acquired strategy to solve other similar problems?
7. In what way was a new experience acquired?

Microgenetic approach aims at an analysis of changes occurring during solving a given problem that takes into account five dimensions of cognitive growth. They include path, rate, breadth, source, and variability (Siegler & Svetina, 2002; Calais, 2008).

- The **path** of change involves the sequence of problem solving attempts performed by children in their way to gaining required competence. It also shows if the change is qualitative or quantitative.
- The **rate** of change concerns the time or amount of experience the children needed to start using a new strategy in a consistent way. It also includes an analysis of the nature of a change - whether it occurred gradually or suddenly.
- The **breadth** of change reflects children’s ability widely to generalize a new approach to other problems and contexts.
- The **source** of change takes into account factors that evoked observed changes.
- The **variability** of change enables evaluation of individual traits of a child in acquiring other dimensions of change. In other words, it enables creating a characteristics of an individual child as well as comparing a pattern of change across individuals (Siegler & Svetina, 2002; Flynn et al. 2006; Calais, 2008)).

It is due to the fact that concentration upon the process of change as it is occurring reveals what mechanisms underpin it. It thus makes possible identifying both detrimental changes...
(areas of dysfunctions) as well as positive ones reflecting a developmental potential of a given child.

5. Microgenesis from neuropsychological point of view

A neuropsychological perspective of the microgenetic theory points to the fact that each action starts at lower levels of the brain and unfolds to the higher more specialized levels. It enables a fresh look upon the nature of symptoms observed in brain dysfunctions. As pointed out by Brown & Pachalska (2003) “the lesion displays phases in a transitional sequence from depth to surface” (p. 4). Two important notions are introduced here: parcelation and heterochrony. Parcelation means the elimination of cells and connections, which occur in over-abundance at birth, in order to achieve specificity. It is connected both with maturation and cognitive experience. Hence, sensory deprivation results in a diffuse and redundant connectivity and a loss of the ability to discriminate among perceived stimuli.

In the case of function the same role as elimination is played by inhibition. Brown and Pachalska state: “Inhibition occurs in the development of action, in newborns, which goes from global movement of the hand or face (the cherubic face of the infant) to one that is more finely individuated” (2003, p. 6). In other words, the basic pattern of each system are elimination, inhibition and specification, which means sculpting away constraints at successive phases of cognitive activity. The authors point out that in pathology re-generalization through disinhibition leads to a number of deviant behaviours.

Another important notion of microgenesis is heterochrony. It assumes that the fact that different brain systems develop at different rates can result not only in adaptations to the environment but also in malfunctions and aberrations. It is connected with the phenomenon of neoteny, which means selective prolongation of an immature phase of development. It makes possible refinement of structures and functions allowing mastering higher cognitive processes of which verbal communication is a good example.

As pointed out by Brown (1998, 2001) microgenesis assumes that phyletic and ontogenetic growth patterns are retraced in microgeny but the processes are collapsed here over a second or in a fraction of a second. Moreover, both mental and motor processes have a hierarchical structure as the later levels unfold out of earlier ones. It is, therefore, possible to analyze the changes in children’s behaviour when they attempt to solve a given problem passing from one level (or stage) to another. And that is of much help in making a course of therapy as effective as possible.

6. Procedure

The aim of our study was to delineate the cognitive abilities of autistic girls in the context of the microgenetic approach. The following question was asked:

What are the characteristic traits of the cognitive development in girls?

In order to find an answer to the above formulated question a detailed description of changing competence of three girls with ASD will be presented. All the three girls represented similar level of autistic features and of cognitive abilities. All of them were able to communicate verbally and all of them were diagnosed in accordance with ICD-10 (International Statistical Classification of Diseases and Related Health Problems) criteria.
The control variables were: depth of autistic deficits, and the level of social, and communicative competence as well as interest in playing and reacting to applied incentives. The elapse of time between individual examinations was also controlled. The boys development was presented in other works (Markiewicz, 2008, 2009; Markiewicz & Pachalska, 2007; Markiewicz & Grochmal, 2008; Markiewicz & Mc Queen, 2008; Markiewicz et al., 2009), therefore their results will be used as a background for the description of girls.

6.1 Case one
Ola B. was first diagnosed at the age of three with the suggestion of middle stage mental disability. No neurological dysfunctions were noted. But her environmental conditions were very bad since her mother died when the girl was 3 months old and her father was an alcoholic. His parental rights were juristically limited when the girl was 2 years old, and she was at the custody of her grandmother (the mother of mother). The first verification of the initial diagnosis was after the girl was 4 years old at our clinic this time. The assessment performed by our team indicated dysfunctions in all spheres of developmental development (F84) suggesting Asperger syndrome (F84.5). The results of psychological examination are presented below.

6.1.1 Results from ICD-10
The evaluation of behaviours performed with ICD-10 revealed:
1. Qualitative impairments in social interactions, which was manifested by:
   a. Difficulties in accepting new situations (e.g. signs of frustration if a sequence of known activities was changed), limited social activity (she did not undertake interactions on her own but undertook simple forms of activity if initiated by an adult)
   b. Weak adaptability to surrounding stimuli, mainly social ones (e.g. she was entirely indifferent to the new persons in her environment)
   c. Emotional distance, lack of spontaneous expression of feelings with the use of speech, gestures, and facial expressions.
2. Communication disabilities revealing in:
   a. Lack of initiating verbal contacts, and limited readiness to communicate
   b. Weak reactions to questions and commands with preserved understanding (she performed simple verbal commands such as ‘come here’)
   c. Weak reactions to visual and/or auditory stimuli, and limited reactivity to non-linguistic messages
   d. Speech limited to simple sentences that often were constructed against grammatical rules
   e. Limited inventory of instrumental gestures and emblems (which commonly are used in place of speech).
3. Behaviour disorders that revealed in:
   a. Many stereotyped reactions (turning round on tiptoes, swinging, beating a floor with an object, and non-functional use of objects)
   b. Lack of correct reaction to stimuli as well as inability to differentiate reactions in response to various character of stimuli, odd treatment of objects, making dices and blocks move.
c. Lack of emotional reactions to new toys characteristic of young children (lack of curiosity and pleasure connected with receiving a toy, interest in new objects)

d. Lack of initiative to start playing.

### 6.1.2 Results from PEP-R (psychoeducational profile – revised)

The scores of Ola on the developmental scale are typical of 30-34 months of age (she was 48 months old at that time). After taking into account emerging scores they rise to 43-47 months of age, which makes it closer to the age of 3 years and 9 months. The highest scores the girl gained in gross motility. At a similar level, yet significantly below her age, were skills of imitation, perception, fine motor, and an ability to come into relationships (relating and effect), sensory responses, and eye-hand coordination. Among them most promising proved to be sensory responses, eye-hand coordination, and cognitive processes with the exclusion of language. Therefore, those three spheres were regarded as the zone of proximal development.

![Graph of obtained and emerging scores of Ola on PEP-R](image)

**Fig. 1.** Obtained and emerging scores of Ola on PEP-R (Psychoeducational Profile – Revised)

Consequent examinations confirmed occurrence of the so called autistic triad, that is trouble getting along with others in a social circle, especially when working together, sharing feelings and thoughts, and making friends; impairments of communication; impairments of flexible imaginative functions of which restricted and repetitive behaviours and interests as well as difficulties in coping with changes are most characteristic. A significant variable, which might have influenced the development and functioning of the girl, was her traumatic babyhood experience. Yet, the character of developmental changes allows the conclusion that it had been rather a secondary factor, though the experiences of her early childhood might have stimulated the appearance of ASD dysfunctions.
6.1.3 Results from WISC-R (Wechsler Intelligence Scale for Children – Revised)
The next examination connected with a need to assess girl’s school readiness at the age of 6:9 revealed a considerable range of scores. Her general IQ was in the range of \(<48 – 76\>\); the scores of verbal IQ were within \(<40 – 58\>; while those of performance scale rose to \(<73 – 98\>\) (\(p = 0.05\)). The highest scores in the verbal scale the girl got in arithmetic – they were within the average. Other scores were at the range of low and very low. In the case of the performance scale it were the block design, object assembly, and coding, which were scored at the average level. Close to average were also her scores on picture completion and picture assembly.

It is also worth pointing out that the girl was able to comprehend simple commands while fulfilling tasks connected with examination, and she also came into verbal contact with the examiner. A three-way analysis revealed that only perceptual organization scored at average level (7.75), resistance to distracters scored below average (5), while scores of comprehension were very low (1).

Thus, the scores gained by the examined girl revealed average abilities in visual analysis and synthesis as well as good perception of abstract stimuli, and good long-term visual memory. At the same time they suggest correct imagination, visual orientation, and visualization as well as a preserved ability to create abstract concepts. Moreover, the scores reflect a good level of planning skills and of simultaneous processing. The girl’s ability to concentrate attention upon a particular task, and an auditory perception of simple stimuli was below average. But most impaired were the abilities to communicate while performing particular tasks. This might suggest that her auditory short-memory was disordered (She was able to repeat only two digits). Auditory perception of complex verbal stimuli, verbal expression, verbal memory, and ability to remember previously learned utterances also proved to be on a very low level.

6.2 Case two
Monika J. was first diagnosed before she was 3 years old. An intellectual retardation of a moderate level (F71) with “some traits of autistic behaviours” was stated at the other clinics. The verification of this diagnosis performed by our team two years later pointed to persuasive developmental disorders (F84), autism in particular (F84.0).

6.2.1 Results from ICD-10
The evaluation of her behaviour in accordance with the criteria of ICD-10 showed:
1. Qualitative impairment of social interactions:
   a. Inadequate evaluation of social and emotional signals, incorrect reactions to the emotional states of others, weak modulation of behaviour in response to a given social context.
   b. Low level of social skills, weak integration of social, emotional, and communicative behaviours
   c. Disturbances in reciprocal social interactions.
2. Restricted, stereotyped, and repetitive interests and activities:
   a. Routine and inflexibility in everyday behaviours, and making others stick to such actions
   b. Stereotyped movements
c. Attachment to the computer appeared with age, including the interest in its construction.

3. Disorders of communication:
   a. Low level of social use of language, weak synchronization and lack of reciprocation in dialogue
   b. Weak changeability of verbal expression; limited ability emotionally to modulate utterances, and weak reactions to questions and instructions as well as to nonverbal clues.
   c. Difficulties in differentiation of rhythm and accent to modulate communication
   d. Limited ability to use facial expressions and gestures in communication.

6.2.2 Results from PEP-R
The scores of PEP-R show that the actual level of the girl’s abilities is typical of the age range 22 – 29 month both in the developmental and behavioural scale. It means that they are significantly below her chronological age as she was 52 months old at the moment of examination. She scored best in gross motor, fine motor, cognitive performance, and perception, while the scores of imitation, language, cognitive verbal, relating and affect as well as play and interest in materials were significantly below average. At the same time, her emerging scores indicated abilities typical of the age range 46 to 51 months so they were close to her chronological age. It was observed in accomplishing such tasks as cognitive performance, imitation, relating and affect as well as play and interest in materials, providing for her zone of proximal development. Scores gained by Monika during an examination with the use of PEP-R are presented in figure 2.

Fig. 2. Obtained and emerging scores of Monika on PEP-R (Psychoeducational Profile – Revised)
6.2.3 Results from WISC-R
The examination administered at the age of 6 years 11 months for the needs of evaluating her school readiness revealed a wide range of scores form moderate mental retardation to an average level. Her general IQ was within the range of <40 - 68>, the verbal IQ was within <34 - 53>, and the performance scores were within the range of <65 – 85>. She accomplished the following subtests of Wechsler Scale on the average level: object assembly, picture arrangement, and block design, while on similarities, vocabulary, and arithmetic subtest she scored very low. It should be pointed out here that despite her difficulties with defining terms the girl was quite good in communicating with others. She was able to understand simple commands and to carry on conversation. She was also surprisingly good at reading messages displayed on a computer screen. She was also able to solve simple puzzles and to put together mixed verses of a poem into a meaningful whole. Moreover, she arranged individual words into sentences, and knew the meaning of road signs. Her arithmetic abilities were also quite good as she recognized and wrote down digits starting from 0 to 100, and she was able to compare digits within the range of ten with the use of signs: =, <, >. Three-way analysis revealed that her perceptual organization was at an average level, while the other factors, such as reasoning (1.5) and resistance to distracters (1) were on a very low level. Her scores suggest average level of visual analysis and synthesis, and of visual perception as well as a correct long-term memory. They also indicate good visual orientation and visualization as well as preserved ability to create abstract concepts. Her abilities to plan and simultaneous processing were also preserved. However, her abilities to concentrate upon a particular task and to interact verbally while performing it as well as her auditory perception of simple stimuli were limited. Very severely disturbed was the perception of complex stimuli, verbal expression, durability of verbal memory, and an ability to remember previously learned expressions.

6.3 Case three
Gabriel R. was first diagnosed when she was 3 years old. It suggested delay and disharmony of her development. The verification of initial diagnosis was done by our team when she reached the age of 4 years and 4 months. It indicated persuasive developmental disorders (F84) suggesting autism (F84.0).

6.3.1 Results from ICD-10
The evaluation of her behaviour performed in accordance with ICD-10 revealed:
1. Impairment of social interactions:
   a. Lack of reciprocal social interactions
   b. Weak modulation of behaviour in response to a given social context
   c. Lack of interest in social aspects of play and/or performing tasks
   d. Lack of spontaneous interactions with the closest.
2. Restricted repertoire of interests and activities:
   a. Sniffing at the surrounding, liking of sharp odours
   b. Stereotyped behaviours, turning round on tiptoes, very quick walking on tiptoes
   c. Obsessive interest in maps and diagrams of technical devices.
3. Decreased communicative competence
   a. Lack of ability to use the language in various social interactions
   b. Inadequate reactions to verbal cues
c. Monotonous non-modulated utterances with agrammatism

d. Difficulties with understanding complex utterances

e. Difficulties with synchronization of linguistic and non-linguistic aspects of communication.

6.3.2 Results from PEP-R

The girl’s scores are at the age range of 37-42 month of life. They are, therefore, below her chronological age. If we take into account emerging scores the age range rises to 50-54 months of life, which means that her potential abilities are close to the chronological age. A detailed analysis reveals that the girl scored best in gross motor, fine motor, cognitive performance, eye-hand coordination, and perception in developmental subscales, while in the case of behavioural scale she scored best on play and interest in materials, and sensory responses. She obtained significantly low scores on language, and relating and affect (behavioural scale) as well as on cognitive verbal, and imitation (developmental scale). The zone of proximal development included relating and affect, language, imitation, and cognitive verbal subscales. The scores of PEP-R test administered at the age of 4 years and 4 months are presented in figure 3.

![Bar chart showing obtained and emerging scores of Gabriel on PEP-R (Psychoeducational Profile - Revised)](image)

Fig. 3. Obtained and emerging scores of Gabriel on PEP-R (Psychoeducational Profile – Revised)

6.3.3 Results from WISC-R

Next stages of diagnostic-therapeutic procedure confirmed the existence of an autistic triad. At the same time, an examination of school readiness at the age of 6 years 11 months...
indicated the distribution of scores within the range from moderate mental retardation to below average. The scores were within <56 – 74> for the verbal scale, <63 – 83> for the performance scale, and <52 – 80> in the case of the full scale IQ (confidence level p = 0.05 ). The highest verbal IQ scores (ranging within average results) the girl gained on arithmetic and information. The remaining scores were very low. As far as the performance IQ is concerned she scored on average level on picture completion, block design, and object assembly subtests. On the remaining subtests her scores were below average (picture arrangement) or very low (coding). She also exhibited difficulty in defining concepts (on vocabulary subtest) in a way similar to the two above described autistic girls. She was, however, quite good at comprehending simple commands and instructions. To give the reader some insight into the nature of the girl’s difficulties we give some samples of her responses appearing in the vocabulary subtest: ‘umbrella’ – like rain; ‘clock’ – bim-bam; ‘alphabet’ – a,b,c…(she made an attempt to enumerate the letters of the alphabet); ‘nail’ – it is cut or not cut; ‘bicycle’ – she lied down on the floor and showed how to ride a bicycle; ‘knife’ – she performed the movements of cutting. Three-way analysis revealed a correct level of development of perceptual organization (7.5) and of resistance to distracters (8.7). Yet, her score on verbal reasoning was very low (2.7). It may reflect her low level of linguistic and communicative competences. On the other hand, the above scores point to good visual processing, and preserved ability to concentrate on a given task. Her abilities to create abstract concepts as well as planning and simultaneous processing were also quite good.

7. An example of a task on developing social interactions

The microgenetic model makes it possible to directly observe the acquisition of cognitive abilities in the process of training. Below we give an example of a task that made possible the evaluation to what extent the child is able to comprehend the visual perspective of both her and of the parent. The girls were to demonstrate to the parent a toy that they had constructed by themselves. The following elements we assessed:
1. The child approaches the parent
2. The child tells the parent: “Look what I have done”
3. The child demonstrates the toy to the parent
4. The child moves her eyes from the toy to the parent
5. The child moves her eyes from the parent to the toy
6. The child demands a commendable: “Do you like it”

During the experiment the examiner and the child were seated at a table, and the parent was seated at another table at the opposite corner of the room. The child was able to see the whole room, while the parent was seated diagonally at the other side. The examiner presented the child a puppet consisting of blocks strung on a stick with a stand. The blocks could be arranged in various configurations so that the shape of the puppet was changed. The girl constructed the puppet, and then the examiner said:
1. Show me what you have done
2. Look at the puppet
3. Do I look at the puppet?
4. Do you like the puppet?
5. Do I like the puppet?
After a short period of joint play, the examiner handed the child the puppet and said: “Show the puppet to your mum/daddy”. The command was accompanied by pointing to the parent with a hand of the examiner. The parents had been instructed to: (1) put their hands over their eyes, (2) sit with their backs to the child. Hence, in order to demonstrate the puppet the child had to approach the parent and make her look at the puppet, for example by pulling her hand and saying “Look what I have done”. The task was evaluated in the following way:

- 0 points - no demonstrating the puppet to the parent
- 1 point - the child approaches the parent but keeps the puppet and looks at it without demonstrating it to the parent
- 2 points - the child demonstrates the puppet to the parent with an appropriate statement.

Altogether six tasks were performed during one session, and the possible maximum score was 12 points. The same pattern of six tasks was then repeated during three consecutive sessions presented at two weeks intervals, and the analysis took into account a total of all scores gained by the examined girls during all sessions.

8. Results

The scores of the girls described in the present chapter are presented in table 1. They show that all of them mastered a particular schema of action. During the first session only one girl approached her parent in the second task but she did not encourage her/him to take interest in the toy. The remaining two girls reached those criteria only in the fifth task. The qualitative analysis, however, will make it possible to delineate the strategy of forming reciprocal social interactions in a task schema by girls with ASD. After the first session the examiner demonstrated the flow diagram. The demonstration was to show the child what she was expected to do. During the second session the girls were shown pictures, in which consecutive stages of actions to be done were presented. The pictures were shown before each task.

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Table 1. Scores obtained by the examined girls in consecutive sessions.

Below we present the scores of boys in accomplishing the above described task (see table 2). Their results from Wechsler Scale were within average and lower than average range. It
means that the intellectual level of the boys corresponded with intellectual abilities of the above described girls. It must be stressed, however, that all boys exhibited aggressive behaviours, such as screams, squeaks, throwing objects, tearing paper, psychomotor agitation, disobedience, and ignoring commands and prohibitions.

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Table 2. Scores obtained by boys in consecutive sessions.

Comparison of scores obtained by girls an boys shows that:
1. Results of the first sessions were similar to those of girls.
2. Difference appeared in the third session since the performance of the boys was much worse than of girls. They did not support the demonstration of the puppet with an appropriate statement.
3. Negativistic and/or aggressive behaviours were much more frequent in boys. They exhibited strong tendency to scream, to squeak, and were apt to break accomplishing the task.

8.1 Microgenetic analysis
The developmental microgenetic model (Siegler, Svetina, 2002; Calais, 2008) makes it possible to take into consideration five mentioned earlier dimensions reflecting the manner of solving a given problem. They are the path, rate, breadth, source, and variability of changes that occurred in consequence of accomplishing a particular task.
1. An analysis of the path revealed that the changes appearing at consecutive stages of performing a task were mainly of quantitative nature: starting from the lack of demonstrating the puppet to the parent in the initial tasks of sessions, then approaching the parent without demonstrating it, and finally demonstration with a verbal message. It allows the conclusion that the change of competence took place there resulting in an ability to come into social interaction with parents, and to understand their point of view. As can be noted in table 1, two of the girls were able to improve their action already in the third task. The girls approached their parents, and made them look at the puppet. It is worth to remind here that PEP-R examination indicated high emerging scores in imitation subscale in all the three girls. Therefore, the above tasks made use of a significant zone of proximal development.
2. An analysis of the rate of change showed that a difference between the primary and consequent strategy of solving a problem came into being only in the second session of
our study two weeks after the first session. However, it was not possible to state if the new strategy appeared gradually or all of a sudden. Stating it would require a more frequent, everyday, repetition of experimental sessions.

3. As far as the breadth of change is concerned it seems that the girls were not able to modify once learned schema of behaviour. They were not acting in a spontaneous way, and did not express any emotional commitment. Perhaps the use of other experimental strategy would enable a deeper insight into the problems encountered by the examined girls.

4. It was also stated that a carefully designed experimental procedure enables forming desired, and at the same time quite complex, actions in autistic girls. In this case the source of change was demonstration of expected behaviours accompanied by verbal as well as nonverbal clues.

5. It should be stressed, however, that the girls acted in an automatic way. They did not express any satisfaction with their work or a will to boast about their success. At the same time, they tended to stick to the once learned schema of action. It allows the conclusion that there was no variability in their behaviour as no personal traits could be noted in their manner of accomplishing the tasks as well.

Yet, it was experimentally stated that an appropriate therapeutic approach makes it possible developing schemas of social interactions. It results in developing independence and a sense of agency so important for creating a sense of identity in a given child. It is also worth to remind that all the three girls had a high level of perceptual organization. And it is connected with simultaneous and global processing of information based upon visual perception, which enables integration of perceived elements and combining them into meaningful wholes. The rate of mental processes as well as eye-hand coordination were also good. They might have difficulties due to the necessity to work under time pressure and their weak resistance to distractors. On the other hand, all of them scored very low on verbal perception tasks, and exhibited difficulties with comprehension of verbal commands and instructions.

It does not need reminding that communication impairment is the main diagnostic feature of autistic triad. Yet, the majority of studies relay heavily upon verbal instructions given to the examined children. The authors seem to forget that autistic children may have difficulties with understanding verbal commands or need some time to process the information included there. Hence, if the scores of sequential processing of abstract nonverbal concepts are within average range, it allows the conclusion that a given child is also able to perform logical operations on verbal material. Especially, if she is able to see cause and effect relationships in everyday situations. Beside communication problems difficulties with performing experimental tasks may also be due to the necessity of combining two types of clues: verbal (instruction) and nonverbal (demonstration). So the actions of an examiner, who intends to explain the rules of a given task, may in effect lead to a confusion as was the case with well known Piaget’s experiments (see Donaldson, 1978). It is also reflected in the phenomenon of “horizontal decaláge”. That is an inconsistency in the tasks healthy children can perform. For instance, they can solve Piaget’s conservation problem on numbers at age six, but they are not able to solve it on mass until age eight, while the ability to perform conservation of weight task appears only at the age of ten (Wortman, Loftus and Marshall, 1988).

Bearing all this in mind we have used a number of repetitions in order to make sure that the child had understood what was required from her in a given experimental situation. The
aim of the above described task was to make it the child understand the visual perspective of another person. While showing her work to the parent the child established a common field of sharing attention. Awareness of the existence of two different points of view is an important indicator of developing social interactions (Meltzoff, 1995; Repacholi & Gopnik, 1997; Gopnik, Meltzoff & Kuhl 1997). A sequence of behaviours used in the above described experiment was to create an awareness of different points of view in autistic children. As pointed out above the girls were much better in performing those tasks than the boys we were examining during a ten year period.

8.2 Possible sources of differences between girls and boys

It is often stressed in autistic literature that the symptoms of ASD exhibited by girls are more difficult to diagnose than those occurring in boys (Attwood, 2007; Kopp et al., 2010; Skuse, 2009). One of the reasons is higher than in boys’ level of social skills, such as an ability to come into social interactions and a style of behaviour in general. Moreover, deviant behaviours of children with ASD influence not only their own development but also the life of the whole family. At the same time, they have impact upon their relations with peers and adults from outside the family. It concerns their attitudes towards such children in particular since autistic children were believed - and often still are – to be dangerous for others due to their bizarre and odd behaviours. Since boys are generally believed to be more aggressive, another significant factor discriminating functioning of girls and boys with ASD may be a difference in educational treatment (Constantino et al., 2009: Jonson-Reid et al., 2010). Beside social learning biological factors may also play an important role. One of them is the testosterone impact upon the foetus (Baron-Cohen et al., 2005).

It may also be worth to point to another factor that might cause differences between symptoms observed in boys and girls. Namely, a well known clinical fact that female brain is less localized than the male brain (Moir and Jessel, 1992). In consequence, brain lesions in women are less disastrous than in men what may be best observed in aphasia recovery. As pointed out earlier, the process of brain development is connected with elimination of unnecessary connections, which leads to refinement of behaviour (see also Kaczmarek, Markiewicz, 2008). It is highly plausible that in the case of autism the selection is delayed, which results in over-abundance of connections, and in disinhibition leading to sensory overload.

It was noted in other works (Kaczmarek, 2003; Kaczmarek, Markiewicz, 2008) that while creating our own image of the world we concentrate upon matters that are important for us and leave out less significant. Therefore, our world image is simplified to a considerable degree, and thanks to it the surroundings seem predictable. And it is that presumed predictability that gives us a feeling of safety. Thus, meeting a mentally ill person makes us feel a bit nervous because we do not know what to expect from him. The autistic child is not able to single out significant stimuli from the non-significant ones, hence her world becomes unpredictable, incomprehensible, and terrifying. It may lead not only to fits of aggression and self-aggression but also to stereotyped repetitive behaviours so characteristic of autism.

9. Conclusion

Our own clinical practice as well as other studies show that early diagnosis and therapeutic procedures connected with it facilitate socialization of children with ASD. Of particular
significance is the individual approach to each autistic child. It enables evaluating not only level of actual skills but also developmental potential of a given child, which in turn improves efficacy of treatment. If we know a zone of proximal development of a given child, we are able to concentrate on the areas in which the child is prone to succeed. Therefore, the microgenetic analysis of the manners in which the child strides to overcome difficulties while solving particular tasks proves to be of great significance. It gives a therapist tools for developing potential abilities of the child, and not to concentrate on her disabilities as it is often the case. Such an individualized and progressive approach increases the efficacy of therapy and gives the child a feeling of success stimulating her to work.

Taking into account developmental profile of a particular child is a necessity since there is a considerable differentiation among autistic persons both in the inter- and intra-individual dimensions. One of them is the difference between the clinical picture of symptoms in boys and girls. Moreover, studies of Constantino and collaborators (2009) revealed subtle difficulties in communicating with others in 20 per cent of families of children with ASD. They were observed mainly in siblings in whom some traits of autism appear about ten times more often than in healthy population. The authors are of the opinion that at least some of specific traits of autistic spectrum may be hereditary. Therefore, they stress the necessity of taking into consideration sex differences while making a diagnosis of autism. Our study has shown that it is possible to develop a schema of action, or a script, in autistic girls. Moreover, the techniques we applied made it possible to analyze a given change, while it was actually happening, and not comparing behavioral patterns from a pre- and post-change as it is often done. Yet, the script remains a rigid unchangeable sequence of actions, while healthy people change it in accordance with the requirements of environment. It was also noted that the autistic symptoms in girls are less pronounced due mainly to their better communicative competence. For that reason their disorders are often neglected despite the fact that their qualitative character is not much different from the disturbances observed in boys. The differences are mainly of a quantitative nature. It is quite probable that refinement of diagnostic methods will lead to better understanding of their problems causing a dramatic increase of the number of autistic girls as was the case with other clinical syndromes.

10. References


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Estimated prevalence rates of autism spectrum disorders (ASDs) have increased at an alarming rate over the past decade; current estimates stand as high as 1 in 110 persons in the population with a higher ratio of affected males to females. In addition to their emotional impact on the affected persons and their family members (in fact, the latter are often unrecognized unaffected â€œpatientsâ€​ themselves), the economic and social impacts of ASDs on society are staggering. Persons with ASDs will need interdisciplinary approaches to complex treatment and life planning, including, but not limited to, special education, speech and language therapy, vocational skills training and rehabilitation, social skills training and cognitive remediation, in addition to pharmacotherapy. The current book highlights some of the recent research on nosology, etiology, and pathophysiology. Additionally, the book touches on the implications of new research for treatment and genetic counseling. Importantly, because the field is advancing rapidly, no book can be considered the final word or finished product; thus, the availability of open access rapid publication is a mechanism that will help to assure that readers remain current and up-to-date.

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