

Diagnostic errors in autism spectrum disorder and their consequences – case studies

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Summary

This article provides an assessment of the problem of diagnostic errors in autism spectrum disorder. The fact that awareness of autism is on the increase has led to the emergence of a growing number of specialists, as well as other people professionally involved in education and the care of children, who have been noticing features of autistic disorders in ever-younger children. On the one hand, this is certainly beneficial in that the level of knowledge about the symptoms of autism spectrum disorder (ASD) is conducive to the identification of children's difficulties; on the other, however, there are concerns that the knowledge possessed by many of those who come into contact with children is often insufficient to diagnose this subtle and changing spectrum of disorders. Neurodevelopmental disorders are especially difficult to assess in small children due to the overlapping symptoms of various disorders. Additionally, periods of intensive development or regression also overlap. Children aged two or three are still too young to exhibit some of the behaviours specific to ASD, and the assessment of speech development and its understanding may be deceptive. The diagnosis of "under observation for possible autism" is often exaggerated. The article presents two case studies concerning diagnostic errors in autism spectrum disorder, their consequences and vital conclusions.

Keywords: autism spectrum disorder, diagnostic errors, case study

"A slight error in the beginning results in a big mistake in the end."

St. Thomas Aquinas

Introduction

Atypical behaviour in some children gives rise to anxiety in parents, paediatricians, teachers and people professionally involved in the care of children, who are often tempted to diagnose the child in terms of suspected autism spectrum disorder (ASD). Various social and environmental factors contribute to a growing number of children manifesting mild developmental irregularities, such as psychomotor hyperactivity,

clumsiness, delays in speech development, behavioural disorders (mainly oppositional defiant) as well as some of the more severe symptoms of ASD [1]. In fact, the number of people with a diagnosis of early childhood autism, Asperger syndrome (ICD-10) or autism spectrum disorders (DSM-5) is growing exponentially. Looking at the statistics concerning children deemed to require special education due to disability in Poland, one cannot help noticing that in the years 2010-2015 there was a sharp increase in the number of pupils with special needs who were diagnosed with autism or Asperger syndrome, i.e. 572 pupils in 2010; 13,299 in 2013 and 21,883 in 2015. This trend is also clearly visible in the case of nursery school children [2]. The literature concerning ASD gives many examples of situations in which a timely diagnosis enables early therapeutic interventions that are more effective than those made later [3]. In fact, various therapeutic programs [4] are initiated even for the youngest children with ASD and children from high risk groups (mainly siblings of children with ASD and children with early difficulties in social communication). Unfortunately, due to some of the symptoms being indistinct at such an early stage in the child's development, early diagnosis may also have negative consequences.

The fact that the awareness of autism is on the increase has led to a growing number of specialists, and other people professionally involved in the process of education and the care of children, noticing features of autistic disorders in ever-younger children. On the one hand, this is certainly beneficial in that the level of knowledge about the symptoms of autism spectrum disorder (ASD) is conducive to the identification of children's difficulties; on the other, however, there are concerns that the knowledge possessed by many of those who come into contact with children is often insufficient to diagnose this subtle and changing spectrum of disorders. In fact, neurodevelopmental disorders are especially difficult to assess in small children due to overlapping symptoms of various disorders. Additionally, periods of intensive development or regression also overlap, and it is often too early for two- or three-year-old children to exhibit some of the behaviours specific to ASD [5].

One of the difficulties with verifying the actual ASD diagnosis is that if it is made by a specialist or a team of specialists it is hardly ever questioned, even though it can be confirmed through longitudinal tests and/or another diagnostic examination made a year or several years after the initial tests. ASD is, after all, considered to be a chronic disorder with a permanent symptomatology [6], though there are also researchers who point to a significant variability of symptoms and varied level of functioning in ASD-diagnosed patients [7, 8].

Kruk-Lasocka [5] emphasizes that the diagnosis of "under observation for possible autism" is usually overblown, especially in cases in which autistic symptoms are weak and the child does not speak. There are examples of 23 children from Lower Silesia found in the period of 2006-2007 who were pre-diagnosed with ASD at the age of 2 to 3 and subsequently recommended for an early intervention program. A year later, following further tests and therapy, the diagnoses were reviewed and childhood autism was confirmed only in four of the children, Asperger syndrome also in four, microdamage of brain function with features of attention deficit hyperactivity disorder

was found in seven, intellectual disability with speech disorders in two, and speech disorders related to developmental dysphasia in two of the children.

Turner and Stone [9] discovered that out of 48 children diagnosed with ASD at age 2, only 63% had the diagnosis confirmed at the age of 4. The children whose diagnosis was unconfirmed displayed lower symptom severity as far as social interaction, communication and limited/stereotyped behaviour were concerned; they also showed better cognitive functioning and more adequate response in various social situations than had been the case during the initial examination. In fact, the stability of diagnosis for children younger than 30 months old was assessed at only 52%. These authors demonstrated a lower stability of diagnosis than was the case in some previous studies [10, 11]. One of their explanations was that the growing awareness and knowledge of ASD made people more sensitive to autism-related symptoms, among them those who were not clinicians but were professionally involved in taking care of small children. They found that these professionals often overinterpreted symptoms and suggested to parents that they suspected their child had ASD. Unfortunately, clinicians have also been found to put forward the ASD diagnosis in cases of children whose symptoms are perhaps not very severe and therefore of uncertain origin.

A review of the research conducted by Helt et al. [12] demonstrates that between 3% and 25% of children previously diagnosed with ASD at some point “lose” the diagnosis. Other studies show that carefully examined children who were considered wrongly diagnosed with ASD were described by their interviewers as timid, fearful, rebellious and not very attentive during the examination [13, 14].

Research carried out in recent years has identified a group of children who reach significant improvement in the area of social contacts, communication, cognitive and adaptive functions to the level expected of children developing normally [15]. In such cases the ASD diagnosis becomes no longer valid. The authors regard early and intensive therapy to be responsible for the change, but other explanations are also possible, such as diagnostic errors [16, 17], effects of a usual maturation process [18, 19] or the fact that parents need the ASD diagnosis to have access to some benefits or specific medical services [20].

Blumberg et al. [21] compared a group of children aged 6-17 with valid ASD diagnoses ($n = 1420$) to those who had it previously ($n = 187$). According to these investigators, the change of diagnosis due to diagnostic error occurs in 13% of children (up to three quarters of the cases were diagnosed on the basis of the information gained from the parents) as well as due to natural maturation (individual developmental path) or therapy (21% of children). In some children the diagnosis was changed (mostly to ADHD), others turned out to be within a normal range.

Compared to children with stable diagnosis, those who have lost their ASD status showed a higher-level functioning and fewer ASD symptoms at the time of the initial diagnosis; it is also the case that their parents expressed significantly fewer worries concerning their children’s verbal skills, non-verbal communication, learning, unusual gestures or movements. Children with a changed diagnosis were also more often diagnosed by non-specialists who were not particularly expert in differentiating between

ASD symptoms and those that are within the norm, transient or characteristic of other health problems.

There is also the issue of the ASD diagnosis being changed to another after a period of time. Fein et al. [19] discuss eight cases of children diagnosed with ASD in early childhood whose symptoms developed and led to a diagnosis of ADHD at school age.

Thus, the following factors might be indicative of a higher probability of a “loss” of ASD diagnosis: a) early confirmation of language or communication skills [22], b) IQ above 70 [22], c) early and more intensive therapeutic intervention [23], and d) subtype of the ASD diagnosis (*Pervasive Developmental Disorder – Not Otherwise Specified* – PDD-NOS) and Asperger syndrome [24].

The case studies

The issue of pro-diagnostic zeal and diagnostic errors, from the perspective of parents who disagree with a teacher’s suggestion to diagnose their child with “observation towards possible ASD” has been of interest to the authors of the present study. The authors have encountered this problem in their own clinical practice. We are aware that individuals whose narratives involve various problems regarding the perception and understanding of the external world discuss them, and at the same time perceive and transform them, from the point of view of their own cognitive structure or personality. However, when they identified several similar, though unrelated, narratives told by different sets of parents, the authors thought that these stories rang true and might indicate the existence of a real problem. This is when they concluded that it would be worthwhile to present the parents’ experience in the form of case studies but also verify the parents’ narratives through medical and psychological examination.

The two case studies that are presented below come from over a dozen cases from clinical practice of the authors. They refer to the consequences of early pro-diagnostic suggestions towards ASD. A case study is a qualitative research method based on idiographic epistemological attitude which strengths mean understanding and describing human individuality. To start with, a researcher has no assumptions or hypothesis but intention to examine a compound phenomenon in its real background. A case study includes reasoning relying on experience in phenomenological paradigm based on subjective experience of a researcher. An observed object is created by subjective interpretation of a researcher. A phenomenological paradigm provides framework for inferential epistemological attitude and the line of reasoning proceeds “from details to generality”. Observation of patient’s behaviour and experience are concluded. In such a meaning it can be treated as a research strategy, in which a patient is intentionally selected. Researchers must remember to draw conclusions with great caution [25].

The investigators’ analysis was based on interviews with parents that included categorized and open questions about the functioning of the family and the child in the form of case studies. The authors have changed some of the personal data to avoid the identification of the respondents.

Case Study 1

The girl whose case is discussed here was 26 months old when the events took place. She is an only child, born from her mother's first pregnancy two weeks before the due date by Caesarean section; the pregnancy was considered normal. She was born 54 cm long, weighed 2800 g and scored 10 on the Apgar scale. Her mother was 38 and her father 46 at the time of birth. For the first two years the child stayed at home with her mother, who was employed in public administration. Her father is a psychologist with a wealth of clinical experience. The girl's paediatrician considered her psychophysical development to be normal; her assessment at 2 years of age was entirely standard. At the age of 2 the child formed sentences, knew a lot of words and was still in diapers (her toilet training was completed at 32 months). The girl required some help with her meals, liked to eat both at home and at the nursery, and ate a varied diet. She liked to play in ways appropriate to her age and could focus on what she was doing when playing. At the time of admission to the nursery, she did not have a lot of experience with other children, but she was happy to communicate with adults. The child was accepted by the nursery at 24 months after an interview with the director. The parents believed that she adapted well, as indicated by the daily care report and information they received from the nursery about their daughter's behaviour. After a few weeks, the parents became anxious as the child began to have nightmares and wake up at night. Eight weeks after admission to the nursery, the girl's mother received a text message from the director asking for a meeting. Both parents attended and were told by the director that she was very concerned about the child's behaviour. She said that the girl knew the rules but did not follow them (did not observe the nursery regulations), would not listen to what the teachers said, did not respond to her name immediately, did not sit still in line during teaching time, brushed her teeth for too long, arranged toys in sequences, refused to tidy up when told to do so, kept spinning when they gave the musical signal designed to end playtime and begin the tidy-up, and that she talked quietly. The director concluded that all this caused her to seriously suspect that the child had Asperger syndrome. She also added that perhaps the parents may have failed to see it, but that she had a "good eye" for such things and founded her opinion on observation and the tests carried out by the psychologist who occasionally visited the nursery and otherwise worked full-time in a specialist autism disorder diagnostic centre. The parents were subsequently advised to see a neurologist at the centre. They were shocked with these conclusions and the interpretation of their daughter's behaviour; they said they felt "crushed". They never saw their daughter spin pointlessly, which is one of the symptoms related to autism. The father of the child did not sleep at all for two nights after receiving the information. The parents, who were previously happy in their marriage and enjoyed parenthood, were sick with worry and took the problem very seriously. In the weeks that followed, they watched their daughter carefully and followed up on what was going on in the nursery. They also asked the director to arrange a meeting with the nursery psychologist to find out what the reasons were for this quick yet serious diagnostic suggestion. Unfortunately, they were told a meeting was impossible "at

least for now". They then asked the nursery to record the child's spinning. Since they were told that it happened a lot, they thought they could get a recording promptly, yet it turned out that there were problems with doing it. After a while, the teachers produced a ten-minute long video, which was thoroughly analysed, and the teachers began working with the child. The parents made various suggestions, and they soon saw the results; the spinning stopped and so did the nightmares. The meeting with the psychologist could only take place two months later. The father wanted to make an appointment in the private centre at which she worked but it only offered full diagnosis, spread across several meetings. Finally, when the meeting did take place, the psychologist had nothing new to say. When she was asked if she knew of any other possible explanations for the child's behaviour, she replied that she was unable to explain it otherwise than in terms of ASD. The father had analysed his daughter's behaviour and told the nursery specialist that the spinning could also be a symptom of a different, less serious, disorder; following DSM-5, he noted, it could also be a simple symptom of adaptation to the new circumstances. He suggested the latter might be possible as the child was the youngest in the group and the age difference between her and the older children was from several months to over a year and six months, which is a lot at such a young age. Towards the end of the conversation, the psychologist apologized to the parents for a rushed and, as it turned out, not fully justified diagnosis of suspected autism spectrum disorder. The child's worrisome behaviour, particularly the spinning, ceased; she made developmental progress and gradually became open to contact with other children. Two months later, the psychologist confirmed that the worrisome behaviour had stopped altogether. When the parents asked her why she had acted so unprofessionally, by making a diagnosis when she could not be sure of it, she replied that, after all, the girl still spoke of herself in the third person, which was fine at her age, but if it continued beyond age 3, the parents would have to consider the problem of functional echolalia. The parents asked why she would worry them if the girl was only 2 years and 6 months old and so much could change within the next six months. Indeed, three or four months later the girl only spoke of herself in the first person. Notably, the nursery director did not seem very happy with the parents' involvement or their comments about the way their child had been handled, and she suggested more than once that they should consider another place for their daughter, although the ASD diagnosis had been abandoned. To make sure the child was healthy, the parents also saw a child neurologist, who excluded the possibility of any autism spectrum disorder. Two years after these events, the girl is developing very well – although she has indeed changed nurseries. On the basis of a detailed interview with the parents, the authors analysed the girl's behaviour at the age of 26 months and referred the parents' narration to the DSM-5 diagnostic criteria for ASD [26]. Criterion A:

1. There are no deficits in social-emotional reciprocity (-)
2. Well-integrated verbal and nonverbal communication, the girl maintains eye contact (-)
3. The child often shares imaginative play, is interested in peers and typically takes part in playing with little cooperation with other children (typical for her age) (-)

Criterion A is excluded.

Criterion B:

1. Stereotyped motor movements – spinning – mostly in circumstances of turning on music indicating the cessation of playing and the beginning of tidying-up time (+/-)
2. Flexible patterns of behaviour, acceptance of changes (-)
3. Various, changeable, flexible interests adapted to social context (-)
4. Appropriate reactivity to sensory input (-)

Only Criterion B1 may be partially fulfilled. It seems crucial that the symptom appeared in specific circumstances (and can be explained with the process of conditioning) and permanently disappeared. The other criteria are excluded. The authors concluded that the problem presented by the nursery was due to faulty classification of the child's behaviour and exclusion of social, family and developmental background (starting nursery too early, significant age gap, scarcity of social experience with other children, lack of interest with parents' knowledge about their daughter). Shortly after the parents' intervention, the child's situation quickly improved and in the new nursery the psychologist has described the girl as well-developing, rather introverted but appropriately initiating social relations with other children.

Case Study 2

In this case, the subjects were the parents of a girl diagnosed with ASD at the age of 5.5. At the time of birth, the mother, an administrative employee in a large company, was 32, and the father, a researcher at a university, was 37. The girl was born from the first and uncomplicated pregnancy; the birth was a natural delivery without complication and the baby was very healthy, scored 10 on the Apgar scale, was 56 cm long and weighed 3500 g. The paediatrician in charge commented on the proper psychophysical development of the child. The girl mastered all developmental milestones (learning to sit, crawl, walk, say her first words, speak in sentences, play; her toilet training was completed at the age of 30 months, etc.) on time. She is an only child and had little social interaction with other children prior to joining the nursery. Since infancy, the child has been sick a lot (mostly with infections). Her mother's parental attitude can be described as overprotective, up to the point of hypochondriasis by proxy towards her daughter. With every infection, the mother was afraid of complications; hence, she took the girl to see a doctor at least several times and always asked for additional tests to make sure her daughter was not seriously ill, but all the tests confirmed that the girl was in good health. In the first year of the child's life, she was cared for by her mother and for the next two years, the father was with her during the day; sometimes the couple used the help of a nanny. The girl began attending nursery at the age of 3.5. From the beginning, she was sick a lot, so she was also often absent from the nursery. On her return, she was strongly reluctant to join in activities and displayed various symptoms of maladaptation. She cried when saying goodbye to her parents, was sad after they left, ate unwillingly, did not want to play with other children or take part in any structured play. One of

the teachers called her a “crybaby”, which made her even more reluctant to attend the nursery. It was perhaps due to frequent illnesses and absences that her adaptation process was exceptionally long. Subsequently, the girl went to stay with her grandmother for the summer holiday and after this longer period of absence she was unhappy to return to the nursery and continued to miss a lot of days. When one of the teachers went on maternity leave, she was temporarily replaced by another; the girl disliked both teachers. With time, signs of better adaptation could be observed: she was more willing to stay at the nursery, had fewer sad and apathetic moments, and found it easier to engage in play with others. Although she was not particularly close to any of the other children, the parents say that at the time she had two good friends from outside of the nursery, though they were never asked about this. In the girl’s second year in the nursery, the teachers said that things were getting better, but seven months in, the head teacher called the mother and asked for a meeting. This is when she revealed that the teachers thought that the girl had Asperger syndrome and should undergo full diagnosis. In the teachers’ opinion, the girl failed to establish relations with other children in the group, did not have friends, often refused to look her interlocutor in the eye, was apathetic, failed to form a bond with the group, reluctantly participated in any shared activity, always played with the same toys, and made brick towers. She ate the same dishes or products all the time and refused trying anything new, often sat at the same table and on the same chair, which she called “hers”, repeated phrases that she heard from others, and did not listen to instructions. The mother was shocked by this information; also, the parents were surprised that they were not asked to come together to discuss such an important matter. They were also not consulted at all about the child’s behaviour at home or in any other non-nursery setting. Their previous conversations with the teachers had not led to any conclusions of this kind, so the couple found it difficult to comprehend the news. In the aftermath of the meeting, the mother suffered from psychosomatic symptoms such as headaches, sleeplessness, shortness of breath with suspicion of a heart attack, and a lowered mood. Within a short time period, they familiarised themselves with the information about autism available from the Internet and books. It was brought to their attention that the language that the teacher used to describe their child was as if taken directly from the websites they came across. They asked again about how justified some of these statements were, as they believed that some of them were not solid enough to classify the various behaviours of their child as symptoms of ASD. However, when they asked for more details, the teacher, who seemed competent, could not really explain the terminology and in the days to follow, the girl did not display most of the behaviour considered characteristic for ASD, e.g. always playing with the same toys. The parents were very anxious and decided to visit a child neurologist, who did not see a need for further diagnostic workup but at the same time did not firmly exclude autism. In the meantime, the girl’s mother started psychotherapy as she suffered from severe anxiety caused by the trauma of suggested ASD in her daughter. The parents also visited a private diagnostic centre where a team of specialists found insufficient grounds for an autism diagnosis (based on ICD-10 diagnostic criteria). The girl’s social relations were assessed as poor (the

relations from non-nursery settings were included). The remaining diagnostic criteria of ASD were unfulfilled (appropriate eye contact, facial expression and gestures, emotional reciprocity, spontaneous sharing of emotions and interests, proper language development and communicativeness, various make-believe play often mimicking observed situations, flexible and age-appropriate patterns of behaviour and interests). The parents were advised of the need to pay attention to the development of their daughter's social interactions. The child continued to attend the nursery without much change in behaviour. However, a lot has changed since she began school. She adapted quickly, likes her teacher and found new friends. Despite of the fact that two years have passed since the misdiagnosis by the nursery teacher, the parents still have strong negative emotions about the whole situation.

Conclusions

The term “diagnosis” is understood broadly as a construct that affects the thinking, feelings and actions of people who make the diagnosis, and also impacts the people on whom the diagnosis is “imposed” and who are directly or indirectly affected by it. Usually, the diagnosis of a child with autism spectrum disorders takes place on three levels:

1. Diagnosis without a diagnosis – at the teaching and pedagogical level, when the “diagnosis” is a result of the teacher's observations and there is interpretation of these observations in psychopathological terms. Sometimes, the observation and its interpretation are made by a psychologist who occasionally visits the nursery.
2. Incomplete nosological diagnosis – based on a diagnostic process that is too selective, early or inaccurate. The diagnosis is considered as final or temporary; this stage also includes the diagnosis of “under observation for possible autism”.
3. Full nosological diagnosis – carried out by a multidisciplinary team, inclusive of psychiatric, neurological, psychological and pedagogical assessments and an evaluation by a speech therapist. This process meets the criteria of a reliable diagnosis (multilateral diagnosis with enough time devoted to it; multidisciplinary, based on multiple observations of the child and a set of confirmed empirical data).

Non-reflective pre-assumptions and actual errors may appear at any diagnostic stage, but most of them take place at the first stage identified above. Although this level of the diagnostic process has no formal or legal implications, it certainly affects the everyday life of the child and the family, the child's functioning at the nursery, and the identity of the family and the child. It also affects the intensity of the stress experienced and may, at this level, be itself traumatic and not unlike the moment of receiving a reliable diagnosis. After all, learning about a child's disorder can be a very traumatic experience [27,28].

These conclusions are largely based on the authors' own interpretation of the case studies and only partly on the literature of the subject. Below are listed some of the non-reflective pre-assumptions that might underlie many diagnostic errors:

- absolutisation of individual observations, ignoring or insufficiently considering other data available about the child;
- insufficient consideration given to the boundaries of one's own actions or the impact of teachers and their beliefs in the mission of taking actions "for the good of the child" (i.e. "the good" as imagined by the teacher rather than the real good as negotiated with the child's parents);
- a pathogenetic approach; focusing attention almost exclusively on the disturbing or pathological aspects of the child's behaviour;
- inadequate attribution of data from the practitioner's own observations to the child's internal characteristics or states;
- impact of "diagnostic fashion", previous training sessions, media coverage (e.g. a lot of coverage given to ADHD and more recently to ASD);
- secondary benefits of diagnosis – financing for the nursery, additional work for teachers;
- insufficient knowledge of the child's clinical and developmental psychology.

Consequently, the errors that appear make it difficult to put forward a reliable diagnosis. These are:

- inability to separate observation from interpretation; the worrying signals in the child's behaviour may be treated dogmatically as the symptoms of a disorder;
- the error of over-radical classification; unjustified application of psychopathological terminology to a specific behaviour of a child;
- insufficient consideration given to the context in which a disturbing behaviour occurs; failure to consider the sequence of events, teacher's own impact on the child, symbolic or metaphorical meaning of a behaviour (e.g. drawing attention to oneself);
- error linked to the pride of educators or doctors; the "We know better", "Parents cannot see as much" approach; labelling the family as "difficult", "parents are in denial", teachers or doctors feeling superior;
- uncertainties being resolved against the child; for example, "spinning" being interpreted as definitely autistic rather than a stereotypic movement disorder or part of the adaptation process;
- disregarding interpretations that are favourable to the child and the family, e.g. adaptation, individual pace of development, psychogenic, contextual or systemic causes; noticing mainly difficulties where the potential of a child should be noted: "she switches herself off" rather than "she can focus on what she is doing";
- iatrogenic errors in passing information to the parents; "something is not quite right with the child, you must see a neurologist", "she has no friends" (about a 3.5-year-old), "its autism, time to diagnose" (after several weeks in the nursery);

- educators crossing the boundaries and acting in excess of their powers when advising parents to see a specialist during a meeting at which they share their observations about the difficulties experienced by the child for the first time;
- projecting their own problems or educational failures onto a child: “she does not comply with the nursery regulations” (about a 2.5-year-old child), “when a group is taken to the toilet, she doesn’t stop what she is doing like other children do, but continues to play” (about a 2.5-year-old child)
- insufficient empathy for the parents’ trauma after the pseudodiagnostic action is taken and failure to consider their need for detailed information; the “diagnostic curse” of a psychologist or a teacher after a selective observation of a child.

With all this in mind, it is advisable to give further consideration to these common errors so as to avoid the consequences they may have for the child and the whole family.

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