

Misdiagnosis of High Function Autism Spectrum Disorders in Adults: An Italian Case Series

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Abstract

High Function (HF) Autism Spectrum Disorders (ASD) in adulthood is highly prevalent but insufficiently recognized. In Italy, in particular, awareness of this condition is still insufficient and many psychiatrists have no cases of HF ASD to mention. Adult patients with HF ASD come to the attention of Mental Health Services complaining of difficulties within their social context and interpersonal relationships.

Objectives: Describe emblematic clinical examples of misdiagnosed HF ASD to understand reasons that conducted to misdiagnosis.

Procedure: We contact five specialized Italian Center in diagnosis of ASD. Each center have to describe two or three emblematic cases of adult patient with diagnosis of ASD validated by ADOS-4 but referred to clinicians with another diagnosis, discussing about possible reasons of misdiagnosis.

Sample and Results: We have collected 12 case reports (2 from Bologna center, 3 from Torino center, 3 from Pavia center, 2 from Verona center and 2 from Catania center) of adult HF ASD previously misdiagnosed. These cases shows important similarity across centers and highlight that if are taken into account only problems or symptoms that conduct patients to ask help, cases can easily suggest other psychiatric or personality disorders. Diagnosis becomes clear only after considering all the clinical features and a detailed developmental history.

Conclusion: Psychiatrists who have insufficient experience of ASD may overlook some symptoms of the overall clinical picture and misdiagnose ASD as personality disorders, schizophrenia, phobia or even as a non-psychiatric condition, so is hopeful for future increased knowledge about HF ASD in adulthood.

Keywords: Autism; Pervasive development disorder; High function; Adulthood; Misdiagnosis

Introduction

Autism is a neurodevelopmental disorder characterized by deficiency in three areas: social interaction, social communication and behavioral flexibility. Using DSM-IV [1], patients could be diagnosed under four separate disorders: autistic disorder, Asperger's disorder, childhood disintegrative disorder, or the catch-all diagnosis of not otherwise specified pervasive developmental disorder, while the DSM-V unified these disorders under a single umbrella, called Autism Spectrum Disorders (ASD).

Epidemiological data demonstrate that the prevalence estimate for ASD has increased over time from 4.4 per 10,000 [2-5] to 62 per 10,000 in global world [6] and can be as high as 100 per 10,000 in England [7,8]. The main explanation for the increased prevalence of this disorder is the notion of Autism as a spectrum rather than a core category condition.

Symptoms and deficit of ASD are differently expressed along the spectrum: peculiar manifestations of the disease can be seen in the most severely disabled patients, whereas other manifestations are seen in patients with a lower degree of disability [9]. The patients with the lowest expression of the disease, 'High Function' ASD and Asperger's Syndrome [10], are the most frequently misdiagnosed for example as schizophrenia [11] or personality disorders [12] because they are more distant from nuclear cognitive and verbal deficits.

Most of our knowledge on ASD derives from clinical and

epidemiological studies in children, only in recent years course of ASD in adults has become focus of new research. A paper reporting on the epidemiology of ASD in adults indicates 1% prevalence in the adult English household population, of who most were, unrecognized [13]. One of the reasons accounting for missed diagnosis may be different clinical features of this disorder in adulthood. Increased prevalence of ASD and high number of unrecognized cases in adulthood cause concern among health institutions. In most countries national guidelines, recommendations and protocols on the correct diagnosis of ASD have been issued. In Italy the National Institute of Health has released a set of guidelines for the diagnosis of Autism and a panel of experts on Autism has issued a set of recommendations suggesting the autistic patient's need for life-long care. Despite these recommendations, in Italy there are only few dedicated centers for diagnosis and treatment of ASD in adults and the problem seems not to be adequately held in most AMHS. The aim of this study is to

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describe some examples of misdiagnosed HF ASD and discuss reasons for delayed recognition.

Methods

Illustrative cases of HF ASD in adults with a history of misdiagnosis were collected by five Italian outpatient AMHS. The centers of Turin, Pavia, Verona and Catania have dedicated services for adult autism, while in Bologna there is an ongoing project in collaboration with Children and Adolescent Mental Health Services (CAMHS).

Evaluation test for ASD

Cases previously misdiagnosed were finally labeled as ASD by a battery of tests:

-Clinical diagnosis: DSM-V criteria for ASD.

-Wechsler Adult Intelligence Scale-Revised (WAIS-R): for evaluation of patients' cognitive level [14].

-Autism Spectrum Quotient (AQ): a questionnaire consisting of fifty questions, it aims to investigate whether adults of average intelligence have symptoms of autism or one of the other autism spectrum conditions [15].

-Empathy Quotient (EQ): a self-report questionnaire for use with adults of normal intelligence that contains 40 empathy items and 20 filler/control items. On each empathy item a person can score 2, 1, or 0, so the EQ has a maximum score of 80 and a minimum of zero. Most people with Asperger's Syndrome or high-functioning Autism score less than 30 [16].

-Autism Diagnostic Observed Schedule, Module-4 (ADOS-4): a semi-structured assessment can be used to evaluate almost anyone suspected of autism and consists of various activities that allow one to observe social and communication behaviors related to the developmental disorders. These activities provide interesting standard contexts in which interaction can occur. The ADOS includes four modules. The individual being evaluated is given just one module, depending on his or her expressive language level and chronological age. Module 4 is used with fluent adolescents and adults [17].

For differential diagnosis with other axis I or of axis II psychiatric disorders we used SCID I [18] and SCID II [19].

Results

We described twelve illustrative cases of patients, with normal

Case #	Intelligence Quotient	Autism Quotient (cut-off >32)	Empathy Quotient (cut-off <30)	Autism Diagnostic Observation Schedule Mod.4 (cut-off autism: 10, cut-off spectrum: 7)
1	73	37	25	12
2	85	35	30	10
3	114	39	23	15
4	126	24	23	13
5	97	38	18	11
6	138	35	19	11
7	137	43	19	10
8	117	37	27	17
9	90	38	32	11
10	107	36	33	10
11	75	35	28	9
12	90	39	29	9

Table 1: Results to tests.

intelligence (IQ>70), only recognized as ASD after correct clinical evaluation in dedicate centers, as summarized in Table 2. This diagnosis was confirmed by the assessment instruments described above. Results of these evaluations are reported in Table 1. As you can observe, all patients stand the cut-off of ADOS-4, gold standard test for diagnosis of autism (10 patients) or for autism spectrum conditions (2 patients). You can also notice that AQ and EQ are negative only in one and three cases respectively. So, as yet know in other research [15,16,20,21]are easy and fast tests, not enough for diagnosis, but useful for screening ASD.

Case report 1 (Bologna)

D. is an 18 year-old boy attending the last year of Technical Institute secondary school, where he has great difficulty both in learning (he's failed one year) and in relating to classmates and teachers.

He has been in care at Bologna CAMHS since he was 5 years old and now, at 18 years old, the Italian health service regulation demands young patients be referred to the AMHS. The CAMHS diagnosis of Behavioral Disorder (F 91) in childhood evolved to one of Avoidant Personality Disorder (F 60.6) in adolescence.

D. appears younger than his age. He looks like a frail child and his body language communicates vulnerability. The WAIS test showed a mild global cognitive deficit but significant difficulties in logic and abstract thought are observed upon clinical examination. D. has great difficulty in dialogue and in expressing his wishes, thoughts and experiences. The tone of voice is low, he has a stereotyped smile, his spontaneous language is poor, he only answers specific questions and with an effort, often after looking at his mother. D. shows a great lack of common sense, often he doesn't get the message because he only focuses on the literal meaning of the words.

The problem most perceived by family members and teachers since childhood has always been difficulty in social interaction. Right from the first year of schooling he showed serious problems of integration in group-class: D. avoided school-friends and had panic symptoms and psychosomatic manifestations when he was due to leave home for school. He was particularly worried about the child throng at school entrance time. Even today, D. becomes extremely anxious when faced with individuals not belonging to the family entourage and has a tendency to isolation. He manages to attend a few group activities (playing guitar and playing judo) decided by his parents in order not let the family down. However, D. perceives these activities as a duty and has never integrated in the group or formed any significant relationships with peers.

D. has a restricted range of interests. His greatest passion is for railways and he spends most of his free time improving a huge plastic railway with meticulously manufactured accessories, a task requiring an inordinate amount of time. His mother reports that D. is extremely rigorous and meticulous and is very good in separating the rubbish collection. D. has difficulty dealing with any change of environment or habits, e.g. a family picnic or a new pet coming, both of which he felt as an undue strain on his mental balance. D. is not conscious of his problems and he is not aware of his future.

D's social interaction difficulties were appraised as the most alarming problem by the family and the school and were the reason why D. was referred to three CAMHS. D.'s good command of grammar and acceptable cognitive level may have contributed to the initial misdiagnosis. But, when we conducted a more in-depth examination of this patient, we noticed some other clinical features like deficits in

Case #	Misdiagnosis	Age at diagnosis of ASD	Symptoms at onset	Possible reasons of misdiagnosis
1	1. Behavioral Disorder 2. Avoidant Personality Disorder	19 years	Difficulties in social interaction, especially in group	Focus only on difficulty in social interaction without integrate this symptom with patient impaired communication and narrow interest, presents since childhood
2	1. Language Disorder 2. Behavioral and Emotional Disorder	20 years	Difficulties in verbal communication and then in social interaction (loneliness)	Focus only on a part of clinical picture (deficit in social interaction) without integrate this symptoms with patient impaired communication and narrow interest
3	Social Phobia	19 years	Difficulties in relating with others, especially at school	No psychiatric history as to the quality of patient relationship with other children had been collected in detail. Nobody observed an unusual way of speaking (logorrhea not related to mood disorder) and difficulty in understanding communication from others, especially in the non-verbal channel.
4	Depression	23 years	Life in loneliness	Focus only on a part of clinical picture (deficit in social interaction) without evaluates restrictive interest and deficit in non-verbal communication.
5	Anxious-depressive symptoms	47 years	Anxious symptoms related to social context	No collect a detail developmental history: difficulties of patient began in childhood relate to a deficit in social cognition. Secondary mental disorder was considered principale symptoms.
6	Anxious Disorder	17 years	Vulnerability to stressful life events and social distress	Secondary mental disorders, as marked traits of apprehension, occasionally manifesting in free-floating-anxiety or anguish with persecutory ideation, were misdiagnosed as the primary disorder but it might be result of deficit in social communication or of her propensity to ritualize and repeat that cause social conflict
7	Personality disorder	50 years	Difficulties in relating with others, especially at work	No in-depth examination of patient psychosocial history from childhood until adulthood. Frequent personal misunderstandings, were interpreted as traits of her personality or as symptoms of different psychological domains. Working independence and certain patterns of exclusion were considered a way to reject relationships whereas represent a way to unload frustration that derives from difficulty in social interaction and social communication
8	1. Borderline Personality Disorder 2. Obsessive-Compulsive Disorder 3. Social Phobia 4. Eating Disorder NOS	23 years	Eating disorder	Many features of clinical picture were in common with other mental disorders, better known by staff and more acceptable to patient's relatives. It seems that a proper differential diagnosis was not possible for two main reasons. First, the hyper-specialization in clinics usually implies poor knowledge of Asperger's Disorder and does not permit a balanced consideration of all the possibilities. Second, ASD high-functioning are probably more clinically confusing.
9	Obsessive-Compulsive Disorder	20 years	Repetitive behaviors	Focus only on one symptoms (repetitive behavior), without check neurodevelopment and without consider lack of social interaction, the delay in speaking and the generally restricted interest like other features of clinical picture
10	1. Language Disorder 2. Behavioural and Emotional Disorder	20 years	Delay in speaking	No integration of symptoms into a single syndrom (social interaction, separation anxiety, delay in speaking and restricted interests)
11	1. Language disorder 2. Behavioral and Emotional Disorder	20 years	Incomprehensible speech and difficults in social interaction	No investigation of the developmental history which shows all three typical features of ASD: deficit in social communication, deficit in social cognition and repetitive and restrictive interests.
12	1. Psychosis NOS	19 years	Difficulties in emotional and relational areas	No investigation of whole developmental history and no integretion of symptoms into a single syndrome, so difficulty in emotional and relational areas and some behavior like watching the washing machine were interpreted as bizarre and collocated under psychotic areas.

Table 2: Features of patients related to misdiagnosis and its possible reasons.

social communication and the presence of restricted, repetitive patterns of behavior, interests and activities. These features, together with the deficit in social interaction, appointed to the more comprehensive and correct diagnosis of ASD.

Case report 2 (Bologna)

V. is a 20 year-old young man. He got his school-leaver's diploma from Technical Institute (secondary school) and has been looking for a job.

He has been in care with our AMHS since he was 18. He had previously been treated at the CAMHS since the age of 4 for Language Disorder (F 80). Speech therapy led to an improvement in verbal communication but during adolescence avoidance of social interaction set in, leading to the diagnosis of Behavioral and Emotional Disorder (F 92) with which he was referred to adult services.

V. has a WAIS-R [14]-tested mild cognitive defect, he looks somewhat 'goofy' and often confused or vacuous. Sometime he

appears rapt in his inner world and not in touch, at other times he keeps murmuring questions before answering them. In the psychiatric interview he displayed unsuitable closeness and was unable to behave as a patient. His utterances were also inappropriate to the subject, talking of his mother's illness in facetious tones, for instance.

There is a lack of social interaction. Since childhood, V. has never had any personal friend or a group to belong to. When younger, he sometimes used to join his brother's group but now avoids going out with them too. V. explicitly says: "I'm not interested in social relationships, because they are not worthwhile and I feel fine by myself". He has never been involved in love affairs. He will say that in childhood he wished to have a girlfriend, but not today. However, in contrast with this emotional gap, V. was very moving when he described his first and only infatuation with a girl: in a matter of seconds, after looking at a girl on the beach for the first time, he fell in love with her and from that moment on him felt 'changed'. His mother reports that V. spends his days at home walking to and fro talking to himself in a low tone of voice. The most frequent expressions that he repeats in an obsessive disturbing manner are: "I'm blessed", "I'm crazy", "I'm a loser". In the same way he will mutter insults addressed to individuals who have hurt his sensibility in the past.

V. has no regrets and rationalizes his lack of friendship and incongruous behavior as inevitable, given his orientation toward rules, whereas feelings and emotions are vain, damaging and to be avoided. His future projects are to the result of rationalizations and focus on buying a car without rear seats, a house in which to live alone, and finding an easy job such as a dustman, without much responsibility and with limited social interaction.

In this patient, loneliness has always been the most worrisome behavioral problem for the family and for the school and this may have caused child psychiatrists to focus only on a part of the clinical picture, thus missing the ASD diagnosis. Only recently have V.'s family understood the importance of impaired communication and narrow interests.

Case report 3 (Turin)

F. is a 19 year-old lad attending the last year of Technical Institute (secondary level).

He did not come to the psychiatrist's notice until last year and had been treated by the AMHS with psychotherapy for social phobia, without improving his behavior. Social phobia was diagnosed because of difficulties in relating with others, especially at school, which started in childhood. Mood disorder and psychotic disorder had been excluded. F. is not conscious of his difficulties and is ego-syntonic in his thoughts and behavior.

At school he shows difficulty in relationship bound up with his deficit in understanding other people's emotions. At the psychiatric examination, F. appeared very rigid in verbal expression, using formal and unusual words. He spoke in a verbose way about the topic introduced. He clearly stated that he didn't understand why people go out at the week-end and why other people feel the need for a partner in life.

He stays alone during the break at school. He has no friends even outside school. The only hobby, as he says, and the only reason to leave his house is going to school.

He needs a rigid organization of space on the table during meals. When he doesn't attend school, he stays alone in his room at his

notebook browsing a fiction website all the time. He has a very sensitive sense of smell and can't stand some foods.

Since his IQ is normal, he was diagnosed as having social phobia, partly because no psychiatric history as to the quality of his relationship with other children had been collected in detail before our evaluation. As distinct from social phobia, we observe an unusual way of speaking (logorrhea not related to mood disorder) and difficulty in understanding communication from others, especially in the non-verbal channel.

Case report 4 (Turin)

B. is a 23 year-old man. Since completing scientific school at the age of 19, he has never worked. He plays piano and paints.

Over the last year he was diagnosed by the AMHS as depressed. No psychotic symptoms have been detected.

He speaks very slowly in a peculiar, stereotyped manner, without mimicry and without modulating his voice. He looks like an old-style English lord, very formal. No eye contact is made.

He stays all day at home with his mother. He spends all his time painting and playing piano alone. He has no girlfriend and no friends, either. Even though his early neurodevelopment proved normal in his medical-psychiatric history, he preferred to stay on his own and has rarely played with other children since childhood.

He likes taking photos but concentrates on a single topic for a prolonged time. Before taking a photo of a little ship made of paper, he first made 15,000 little paper ships, spending several months concentrating on that topic alone. This is his usual manner of organizing his life.

As distinct from depressive disorder, one notes that normal interests are present and he feels pleasure in playing and painting. On the other hand, he concentrates on a single topic to an unusual degree. His way of living alone is not related to negative or positive psychotic symptoms and started in childhood.

Case report 5 (Turin)

M. is a 47 year-old woman. She works as a clerk and has a degree. She arrived at the AMHS with anxious-depressive symptoms to do with difficulties in her job. Since adolescence she has been diagnosed as suffering from anxiety disorder and been treated with antidepressants and psychotherapy. She is clearly ego-syntonic in her behavior and seems to be living in a novel, exhibiting childish behavior.

She says that she has difficulty in social communication. She doesn't understand why recently her boss punished her for a serious mistake but she thinks it is not as important as the director claimed. She explained that she failed to tell her boss about a project and referred directly to the general manager because she didn't understand the implicit instruction behind her boss's remarks. Another time, she lost an important office document on the train and a member of the public found it and brought the document to her boss; she couldn't understand why he was so angry "because in the end the document was found". She had the same problem communicating with colleagues in other jobs and has had to change placements several times on account of her behavior, but she has no difficulty in winning competitive selections, especially when written, and always finds another job. She doesn't understand when it is her turn in a phone call and since childhood she has tended to be long-winded.

She is not able to work with others. Since adolescence she has

shown serious difficulty in making friends at school and outside school. Since childhood she has been “shy” and unable to understand emotions expressed by others.

She needs an orderly plan for her life and prefers to wear clothes of the same color and tactile feel.

The clinical picture can hardly be anxiety disorder alone. The difficulties that began in childhood relate to a deficit in social cognition. In contrast to her IQ, she exhibits some very childish behavior that is not related to a personality disorder.

Case report 6 (Pavia)

E. was full-term born in normal circumstances, and was bottle-fed. He is at present an undergraduate of Psychology at a French university, providing special support for people with autism spectrum disorders. He is in care with a psychotherapist and has periodic checks at our clinic in Pavia.

E. was first diagnosed as suffering from Asperger’s syndrome when he was 17, and was 18 when he first contacted the Autism Lab four years ago.

At kindergarten E. was described as an isolated child, with a lack of interest in play activities.

Of note, E’s speech and thought seem to be unaffected by autistic spectrum disorder, but – upon in-depth examination– one observes an undoubted attention to detail and some other minor oddities. Again, mild ‘dysprosody’ is sometimes detectable, with a tendency to poorly modulate the voice tone.

He has presented strange and bizarre behavior at times during his life. For instance, he might get up in the middle of a meal to remove cheese crusts or other unpleasant food from other’s dishes. In spite of the undoubted improvements achieved, even today E. finds it difficult to understand the motivations underlying others’ emotions, and in the same way he feels a certain embarrassment in facing up to his own feelings. Loud noises, especially if sudden, have been an important issue, as they have triggered panic attacks on many occasions.

His mother reports normal psychic, psychomotor and speech development. On the other hand, E. has presented some peculiarities since he was young, such as a vivid interest in the ‘evil’ characters of films, and a strong, ambivalent passion for high voltage cables: he has repeatedly tried to get close to them, despite the intense fear they cause him.

These peculiar features were never taken into account by the patient or his family as a symptom of ASD and had never been evaluated by psychiatry. Inverse vulnerability to stressful life events and social distress were perceived as a huge problem without any understanding of how it might be a result of deficit in social communication. Hence, secondary mental disorders, which in E.’s clinical picture include some marked traits of apprehension, occasionally manifesting in free-floating-anxiety but also as anguish with persecutory ideation, were misdiagnosed as the primary disorder.

In particular, it is not always possible to separate certain “neurotic” features of E. from family and social conflict deriving from his propensity to ritualize and repeat, which is typical of ASD.

Case report 7 (Pavia)

F. is a woman of fifty and was the editor of two magazines when she arrived at the Pavia Autism Lab for consultation.

In childhood she was quiet and shy; she remembers having been a remarkable student during elementary school, when her teacher protected her. By contrast, over the following years she experienced exclusion and bullying by her classmates. She had a single, partly sentimental/friendly relationship at the age of 19 with a neighbor of hers of the same age. This story had a tragic ending, because the young man disappeared while hiking in the mountains. His remains were retrieved only 9 years later. After his disappearance, F. took a trip to India – the center of interests she shared with the boy – hoping to find him there. Back home, F. got various jobs, and meanwhile had a complex academic career, beginning her studies in Law and then moving first to Philosophy and then to a Naturopathy school. She has unusual interests in nutrition, Hinduism and theosophy. Her first employments were complicated by tensions in her working relationships (mobbing colleagues, incomprehension with staff/superiors). Nowadays she is editor in chief – and the only author – of two magazines dealing with nutrition.

During our first interview, F. acknowledged her difficulty in “reading others’ minds”: she reports constant difficulty in relationships and exchanges with other people. Nevertheless, her speech is rich and fluent.

F. claims that she didn’t suffer much from social exclusion during her youth, because she longed for solitude and had no great interest in making friends or taking part in recreational group activities. Despite the experience she has acquired and a remarkable degree of social success, the patient shows an inability to tune herself with others (both in expressing her own feelings and in perceiving others’ emotions). However, this problem didn’t prevent her from meeting a man who shares some of her characteristics. They married in secret and live in two different towns, but they meet frequently and enjoy mutual trust.

F. asked for a consultation in her fifties and appeared relieved to learn she has Autism Spectrum Disorder. Correct diagnosis was only possible after in-depth examination of her psychosocial history from childhood until adulthood. Her previous life had been marked by frequent personal misunderstandings, which were interpreted by psychiatrists as traits of her personality or as symptoms of different psychological domains. In the past, her working independence and certain patterns of exclusion were considered a way to reject relationships whereas they represent a way to unload frustration that derives from her difficulty in social interaction and social communication.

Case report 8 (Pavia)

F. is a 23 year-old young woman. She was born at full term, after a normal pregnancy, and breast-fed until the age of 2.

F. was a clever pupil, with an excellent academic record, though she learned “much more from books than from teachers”. She took her final high school examination at the same period as her parents’ separation.

After spending her first university year without taking exams, her situation was outlined to a distant relative (a psychiatrist), with whom she had some interviews. Undiagnosed by him, after some episodes of binge eating followed by vomiting and self-injurious acts (forearm lesions), F. was redirected to a Bulimia/Anorexia clinic, with little result: she performed serious self-harming acts by taking drugs (Emergency Department access for attempted suicide in 2009). On being referred to a CAMHS, F. was discharged to an AMHS for further investigation. Finding hospitalization unbearable, F. self-inflicted a deep cut on her arm, involving the tendon. She was then diagnosed with Borderline

Personality Disorder, Obsessive-Compulsive Disorder, Social Phobia and Eating Disorder NOS. F. was prescribed antipsychotic and antidepressant drugs and began psychotherapy. She had no benefit from these prescriptions and she attempted suicide one more time.

When we analyzed F.'s history since childhood, many difficulties in social interaction and social communication became visible: she started preschool at 3, with problems arising from the first day, as F. did not look for company and did not play with schoolmates. From childhood on, she has been in contact with only one friend. She had never tolerated physical contact, often appearing impolite, cold and detached to other people. Beside this, F. has never understood implied gestures and innuendos such as winking, and started to decode facial expressions thanks to her mother's help and watching cartoons.

Admitted to University classes, her efforts to attend lessons were undermined because of her preference for avoiding physical contact (she needed an empty space around her, to be close to the exit, etc...).

F. got in touch with an Asperger's Association thanks to her personal research on the Internet; from there she was redirected to the Autism Lab. She was always marked down as a shy, odd person, but none of the professionals whom she contacted diagnosed her condition properly. Many features of her clinical picture were in common with other mental disorders, better known by staff and more acceptable to her relatives. It seems that a proper differential diagnosis was not possible for two main reasons. First, the hyper-specialization in clinics usually implies poor knowledge of Asperger's Disorder and does not permit a balanced consideration of all the possibilities. Second, high-functioning people like F. are probably more clinically confusing.

Case report 9 (Catania)

G. is a 22 year-old man. He completed his schooling at a Professional Institute. He lives at home with his family (mother, father and a brother) and is not looking for a job. He has been in care at the AMHS since 2012, and was never assessed for developmental disorder during childhood and adolescence even though he only started to talk at the age of 4.

He has difficulty in following a conversation, in particular when abstract thinking is the subject, and sometimes appears to follow his own thoughts instead of answering the questions of the physician, preferring to talk about the weather, a topic on which he is really competent. He manifests a lot of anxiety and tends not to keep eye contact with the investigator.

Motor behavior observation shows movements to be slow and clumsy.

From the collection of his psychosocial history, it emerged that he had difficulty in social relationship at school and at home, where he always seemed really shy and never made good friends. In adolescence, he had no group of friends and no sentimental relationships.

His parents say that he spends most of the time at home, studying satellite maps, talking to himself, discussing meteorological problems. Often he has to count down from 100 to 0 to calm anxiety and if he is interrupted he has to start again and manifests anger. He doesn't want to eat "red food" and won't explain why. Mother also says that sometimes "he talks without thinking about other's feelings".

He has a plan for the future "to become a meteorologist, possibly living in an orbiting station", but he is unable to outline any strategies to achieve this goal. G. was evaluated for the first time when he was

an adult and he was classified as suffering from Obsessive Compulsive Disorder. This diagnosis seems to be a consequence of narrow focusing on repetitive behavior, but when we checked the neurodevelopment and considered the lack of social interaction, the delay in speaking and the generally restricted interest, a clear diagnosis of ASD arose.

Case report 10 (Catania)

D. is a young man of 20, being treated at the CAMHS unit for Language Disorder and referred at 18 to an AMHS with a diagnosis of Behavioral and Emotional Disorder. He started to talk at the age of 5 and went to school for the first time at the age of 6 because he didn't want to leave his parents. He attended school until the age of 18, completing higher studies. At the moment he has no job and is looking for one but is not able to sustain a job interview.

During the collection of his medical history he started describing his life from the first day he remembers and then day by day with a lot of particulars and descriptions and it was really difficult to make him desist. The investigator could only question him when he allowed him to do so; otherwise he would stall with "the subject is exhausted". Speech is not fluent and prosody is more or less absent. Mimic movements are not congruous with the topic or affective status.

He explains that at the moment he is very sad because he feels different from others and is pained at the lack of social relationships; in particular, he would like to have sexual relationships with girls but doesn't understand why they don't want to have sex with him when he asks to as plainly as he knows how.

He has a particular interest in watching cartoons and reading comics, which he relates in a lot of detail.

He doesn't want to leave home, which would mean changing his room and habits.

At the end of the interview he left the room ignoring the physician's outstretched hand. At the time of diagnosis, attention focused on social interaction, separation anxiety, delay in speaking and restricted interests, but there was no integration of symptoms into a single syndrome. This led to misdiagnosis.

Case report 11 (Verona)

B.A. is now twenty. He has got a professional degree in agriculture, thus completing his educational curriculum with the help of a support teacher all the way through. He works as a warehouseman in a local general hospital. He had his first contact with a child psychiatrist at the age of two and received a diagnosis of Language Disorder (ICD10 F80). When referred to the adult psychiatric service he was given a diagnosis of Behavioral and Emotional Disorder (F 92).

As a child, B.A. had severely disturbed, practically incomprehensible speech; he was then treated by a speech therapist between the ages of four and a half to six, with satisfactory results. However, for years to come he would maintain peculiar stereotyped, repetitive verbal sequences as found in animated cartoons, and disturbances in the domain of semantics and pragmatics. These last are still present, together with awkward, emphatic prosody, an evasive attitude when faced with challenging demands, and verbose speech on a limited set of topics.

B.A. is visibly uneasy in social situations and extremely slow in becoming acquainted with anybody. Even today he reports that he feels different from other people in general and that he is content with his "six-friend group".

B.A. is fairly independent in moving around, but he has a limited range of interests and prefers few, repetitive activities. He has cognitive skills at the bottom of the normal range, with restricted logical-mathematical intelligence, and a rigid superficial way of thinking that tends to drift under environmental pressure. He writes in lower-case block letters, with repetitive dashes. When outside, he sometimes feels grabbed by women and by people attending work activities. He shows only partial awareness of his condition and does not seem to have any plans for the future.

B.A. was initially assessed by a child psychiatrist at an early age when his speech disturbance was the foremost symptom; later on, he was not seen by a child psychiatrist on a regular basis and the diagnosis stuck. The diagnosis he received in adulthood is emblematic, because difficulties in social situations, which appear in adolescence, often look like disturbed emotions and behavior. These symptoms are more easily reported to psychiatrists and diagnosed as such without a thorough investigation of the developmental history which shows all three typical features of ASD: deficit in social communication, deficit in social cognition and repetitive and restrictive interests.

Case report 12 (Verona)

M.G. is 19 years old attending the final year of secondary school with a focus on modern languages, in which he has good results. He has been examined by child psychiatrists since he was three with an initial diagnosis of Psychosis NOS. At the time of his referral to the adult psychiatric service he received a diagnosis of PDD NOS with Psychotic Traits.

M.G. is well built, a little clumsy, with relatively poor facial mimicry and a tendency to avoid visual, let alone physical, contact.

He shows good cognitive performance and meets environmental demands fairly well, especially when prompted, but at the cost of some emotional arousal betrayed by either signs of free anxiety or obsessive rituals. He tries hard to keep events under control because he finds it hard to tolerate unexpected changes.

M.G.'s verbal communication is good, but sparingly used, unless prompted. His vocabulary is adequate, but filled with clichés. Social interaction has been poor since childhood: parents recall he was a very quiet child, lacking initiative, with a tendency to isolate himself. They started to worry when they found him standing still in front of the washing machine, staring at, and fascinated by, the revolving drum.

To this day, he continues to experience major difficulties in his emotional and relational areas: he is unable to express his needs successfully so that he is exposed to frustration and tends to blame others for his disappointment. He emotionally distances himself from others so that he has a very limited number of strictly selected contacts. At times he experiences aggressive urges.

He has attended school regularly and also obtained good grades. He is especially fond of reading English newspapers, favoring classified ads and commercials, rather than articles.

He was for years a boy scout; now he attends gym under the supervision of a personal trainer, with good results. In spite of the fact he is finishing secondary school, he does not express any plans for the future; yet, if questioned, he becomes vague and looks distressed. He seems unaware of his condition.

The reason for the original misdiagnosis may be attributed to persistent misinformation about ASD in the medical profession

until recent times. Good cognitive performance and good verbal communication tend to put psychiatry off a correct diagnosis. Difficulty in emotional and relational areas and some behavior like watching the washing machine were interpreted as bizarre and collocated under psychotic areas. However, when we considered the whole developmental history of patient, we came to see all the problematic areas as a single syndrome. A proper understanding of ASD led us to a new reading of the patient's problems and formulation of a correct diagnosis.

Discussion

The aim of this paper is educational. Some misdiagnosed cases of ASD have been described in order to highlight the possible reasons causing child and adult psychiatrists not to recognize cases of HF ASD and to classify them under other psychiatric diagnoses. Patients were finally correctly recognized in dedicated centers and the diagnosis was confirmed using psychometric instruments catering for ASD.

In the Italian Mental Health Service knowledge about HF ASD in adulthood is still far from widespread and the standard of care for this disorder seems to be poorer than for schizophrenia or other major mental disorders. Most Italian mental health service professionals have no cases of HF ASD in adulthood to mention.

The dearth of cases in Adult Mental Health Services is possibly due to the negative illness behavior that characterizes ASD: generally these patients do not seek help, have no awareness of illness, tend to isolation and consider treatment as intolerable intrusion. Another possible explanation is misdiagnosis of HF ASD in adults. These patients are often considered as subjects with social problems or are given a psychiatric diagnosis of the kind more routinely used by psychiatrists.

Again, the literature confirms that HF ASD may not be recognized or may be misdiagnosed as depression, personality disorder, or psychotic illness [11,22,23]. Correct identification of patients with classic nuclear autism is fairly easy because the features of the disorder are well-defined, severe and self-evident. The clinical picture of HF ASD is quite different in patients with indefinite/ambiguous features of the disorder which can only be recognized as Autism if one assumes such a disease to be placed within a spectrum. Recent studies demonstrate that HF ASD represent the extreme end of a normal distribution of autistic-like traits [24], in a continuum from normal to pathological, with different levels of gravity.

The cases described share important common clinical features: all are characterized by normal verbal communication and fluent grammar and all were brought to the attention of a physician because of relational problems, perceived not by the patient but by his/her family members or school teachers. Behaviors concerning interpersonal communication difficulties and patterns of repetitive behavior are less commonly perceived by the patient and his environment as a good reason to visit a doctor.

The specialist evaluating the patients, in turn, may underestimate the problem due to limited knowledge of HF ASD and reluctance to formulate a diagnosis of ASD, for fear of the 'stigma' this diagnosis may involve.

In these subjects, the diagnosis becomes clear only after considering all the clinical features and a detailed developmental history. If only single clusters of symptoms are taken into account, cases can easily suggest other psychiatric or personality disorders which are more familiar to the psychiatrist. Thus, aberration in language and apparently

'bizarre' behavior can induce a misdiagnosis of schizophrenia [11,25-27]; a pattern of repetitive thoughts and behavior can lead to a misdiagnosis of obsessive disorder [28-30]; social withdrawal and difficulty in relationship with others can induce a misdiagnosis of social phobia [31] or schizoid/schizotypal personality disorder [32,33]; poor emotional control can cause a misdiagnosis of borderline personality disorder [34], and so on.

Conclusion

The history of patients described above, shows how difficult may be to correctly identify HF ASD in adolescents and adults and evidence that individuals of normal intelligence with ASD tend to be diagnosed with ASD late in childhood or sometimes in adulthood, despite a persistent symptomatology [35].

In our case series patients received a misdiagnosis both by CAMHS and by AMHS until they came to the attention of dedicated centers for ASD. But only a correct and prompt recognition of these disorders will allow appropriate support for these patients and their families [17]. The effect of inadequate identification and assessment of patients with ASD leads to inadequate or even damaging care [22]. For these reasons, family associations and health service authorities are greatly concerned about this issue, as demonstrated by recent recommendations for recognition, diagnosis and management of adults with autism published by the National Institute for Health and Clinical Excellence (<http://guidance.nice.org.uk/CG142>).

The hope for the future is that proper training in the identification and assessment of autism should figure more prominently in the undergraduate and postgraduate education of health and social care professionals [22].

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