

Sociodemographic and social barriers to early detection of autism

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ABSTRACT

Background. The increasing incidence of autism spectrum disorder (ASD) is a common finding of many studies. Early diagnosis and appropriate treatment approaches for ASD can provide favourable clinical outcomes. This study aimed to determine the factors affecting the age at diagnosis, in children with ASD.

Methods. Two hundred and two cases diagnosed with ASD were included in the study, according to the DSM-5 diagnostic criteria, at the Mersin City Training and Research Hospital Child and Adolescent Psychiatry outpatient clinics, between April 2021 and August 2022. Clinical features and sociodemographic data that may be related to early diagnosis were investigated.

Results. The mean age at diagnosis was 36.76 ± 15.30 months. In 71.3% of cases parents were the first to suspect that children were developmentally different. In 38.1% of the cases, at least one of the parents denied the symptoms and evaluated their child's development as age-appropriate. It was found that 32.7% of the cases evaluated by pediatricians and 32.5% of cases evaluated by family physicians, were referred to child psychiatry examination. The present study revealed that higher educational level of the father and the middle-high socioeconomic status, were associated with early diagnosis. There was also a positive correlation between paternal age and age at diagnosis.

Conclusions. The age at diagnosis is below the target level for early diagnosis. Studies should focus on increasing awareness of health professionals and parents about ASD.

Key words: autism, early diagnosis, sociodemographic factors, awareness.

Autism spectrum disorder (ASD) refers to a group of differences that typically appear in early childhood and are characterized by repetitive and restricted patterns in behaviours, interests and activities, as well as difficulties with social interaction and communication.¹ It's reported that the incidence of ASD is increasing all over the world both within society and in the health community, mainly as a result of an increase in awareness of the disorder, increase in the number of diagnostic centers and changes in diagnostic practices, although the increasing

paternal age and other factors have not yet been adequately clarified.^{2,3} According to the Center for Disease Control and Prevention (CDC), the prevalence of ASD was reported to be 1/160, 1/68, and 1/59 in 2000, 2014, and 2018, respectively, in the United States, and a striking increase in the incidence of ASD has been revealed.⁴ This increase in incidence rates shows the high probability of all healthcare professionals, especially physicians, of encountering children with ASD.

The most important issue emphasized in the literature in terms of the prognosis of the disorder is the early diagnosis and the initiation of appropriate educational approaches, without delay.⁵⁻⁷ The main goals of special education practices that are used effectively in the

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treatment of ASD, are to reduce core symptoms and increase adaptation skills, in order to enable the individual to reach the upper limit of their own potential. Results from extensive clinical experience show that families begin to notice ASD symptoms after the age of one.

However, in studies conducted in Türkiye, it's known that the age at diagnosis is still far behind the target. The inability to reach early intervention due to delayed diagnosis has a direct negative impact on the clinical severity of the disorder. Although there is no fully accepted time frame for early diagnosis in the literature, 3 years of age is accepted as the age limit for early diagnosis in studies investigating various factors related to early diagnosis and the clinical importance of early intervention.⁸⁻¹⁰

A number of studies have been conducted for early diagnosis, both in Türkiye and in other developed countries, in the hope that the age of diagnosis may be reduced to the lowest level possible.^{11,12} In addition to family-related social variables, quality and access to health care are also important for age at diagnosis. Families in our country are evaluated by various health personnel, primarily by pediatricians and family physicians, until they apply to Child and Adolescent Psychiatry Clinics where they are diagnosed with ASD. The rising incidence of the disorder and the ease of access to health services both increase the possibility for an encounter between these patients and health professionals. However, the most significant reason the targeted age for early diagnosis has not yet been reached is the lack of sufficient awareness regarding the issue. Consequently, increasing awareness in healthcare professionals who are likely to encounter children with ASD, will assist to achieve this goal.

In this study, it was aimed to investigate the age of diagnosis and the variables thought to be effective in the diagnosis process. Another aim of the study was to contribute to the early diagnosis of the disorder by raising the awareness among healthcare professionals,

who are especially likely to encounter toddlers and preschool children.

Material and Methods

This cross-sectional study was conducted in Mersin City Training and Research Hospital Child and Adolescent Psychiatry Unit, between April 2021 and August 2022. Cases diagnosed with ASD for the first time according to the DSM-5 diagnostic criteria were included in the study. Those who had previously been diagnosed with ASD in another center, those with neurometabolic disease and genetic syndromes, were excluded from the study. The diagnosis of ASD was made after two or more consecutive clinical interviews, and the patients were re-evaluated by a second child and adolescent psychiatrist to confirm the diagnosis.

Sociodemographic and clinic information forms were completed, where the age and gender of the child, concomitant diseases, birth history, family's complaints, family structure, age and educational status of the parents, and previous health care institution applications of the family were collected. The Hollingshead-Redlich Scale was used to determine the socioeconomic status of the family, whereby parents were divided into three groups as low, middle, and high. The health facility applications of the patients in the last six months were controlled via electronic information systems, with the approval of the family.

Statistical analyses were performed using the SPSS Statistics for Windows, version 26.0. Variables were tested for normal distribution using the Kolmogorov-Smirnov test. The demographic variables were expressed as mean \pm standard deviation, number (n), and percentage (%). The relationship between variables was assessed using Spearman correlation analysis. The Pearson chi-square test or Fisher's exact test were used to compare categorical variables. A p value of less than 0.05 indicated a statistically significant difference.

The approval of the Clinical Research and Ethics Committee of the Mersin City Training and Research Hospital were obtained for the study (03.03.2021/194). Written consents were obtained after detailed information was given about the subject and purpose of the study.

Results

Two hundred and two cases diagnosed with ASD were included in the study; the sociodemographic data of the participants are summarized in Table I. The mean age at diagnosis was 36.76 ± 15.30 months. Of the participants, 165 (81.7%) were male; the clinical characteristics of the cases are summarized in Table II. While mothers came to the interview alone in 114 cases (56.4%), mothers and fathers came together in 88 cases (42.6%). During the diagnostic interviews, the most common reason for referral was speech delay. The complaints of the families at the time of application and their frequency are provided in Table III.

The cases were divided into two groups, as those with an early diagnosis and those with late diagnosis, the cut-off was selected as 36 months. Determination of this cut-off point was based on studies in which the age of early diagnosis was defined and it was also aimed to distribute the number of patients into the two groups in a similar fashion: 117 of the cases (57.9%) were included in the early diagnosis group, while 85 cases (42.1%) were included in the late diagnosis group.

The results of our research reveal that the educational status of the father is associated with early diagnosis. The children of fathers with a secondary education level and below, were diagnosed later than the children of fathers with a higher education level. In addition, it was found that cases in which at least one of their parents denied their symptoms, were diagnosed later than those whose parents did not deny their symptoms. Again, children whose symptoms were noticed for the first time by third parties other than their parents, were also diagnosed

late. Early diagnosis was significantly higher in the children of families with a medium-high socioeconomic level, compared to those with a low socioeconomic level. There was no relationship between the gender of the child and the educational status of the mother and early diagnosis. It was determined that variables such as epilepsy, preterm birth, presence of regression in the history and family structure, were not associated with age at diagnosis. Being evaluated by a pediatrician or family doctor in the last six months was also not associated with early diagnosis (Table IV).

Table I. Sociodemographic characteristics of children (n=202) diagnosed with autism spectrum disorder (ASD).

Age of diagnosis (months)	36.76 \pm 15.30
Gender	
Male	165 (81.7%)
Female	37 (18.3%)
Maternal age (years)	30.60 \pm 5.84
Paternal age (years)	35.20 \pm 6.43
Mother's education level	
Primary school and below	86 (42.6%)
Middle school	51 (25.2%)
High school	45 (22.3%)
University	20 (9.9%)
Father's education level	
Primary school and below	47 (23.3%)
Middle school	45 (22.3%)
High school	78 (38.6%)
University	32 (15.8%)
Parental status	
Married	189 (93.6%)
Separated/ Divorced	13 (6.4%)
Family structure	
Nuclear family	166 (82.2%)
Extended family	36 (17.8%)
Caregiver	
Mother	190 (94.0%)
Others	12 (6.0%)
Socioeconomic status	
Low	118 (58.4%)
Medium/ High	84 (41.6%)

Table II. Clinical and diagnostic features of the cases (n=202).

Who came to the diagnostic interviews?	
Mother	114 (56.4%)
Father	2 (1.0%)
Both	86 (42.6%)
Who referred family to the child and adolescent psychiatry clinic?	
Families' decision	118 (58.4%)
Pediatrician	33 (16.3%)
Family physician	28 (13.9%)
Other	23 (11.4%)
Who was the first person to suspect a developmental difference?	
Mother	126 (62.4%)
Father	18 (8.9%)
Other	58 (28.7%)
Has the patient been seen by a pediatrician in the last 6 months?	
Yes	165 (82.7%)
No	37 (17.3%)
Has a child psychiatry evaluation been recommended by the pediatrician?	
Yes	54 (32.7%)
No	111 (67.3%)
Has the patient been seen by the family physician in the last 6 months?	
Yes	151 (74.8%)
No	51 (25.2%)
Has a child psychiatry evaluation been recommended by the family physician?	
Yes	49 (32.5%)
No	102 (67.5%)
History of epilepsy	
Yes	11 (5.4%)
No	191 (94.6%)
History of premature birth	
Yes	23 (11.4%)
No	179 (88.6%)
History of regression	
Yes	47 (23.3%)
No	155 (76.7%)
Do parents (mother and/or father) have symptom denial?	
Yes	77 (38.1%)
No	125 (61.9%)

Table III. Complaints and its frequency in children diagnosed with ASD at time of application.

Speech delay	83.6%
Hyperactivity	43.0%
Inability to establish appropriate social relations with their peers	34.1%
Not responding when called by name	32.6%
Not taking instructions	24.7%
Being strictly attached to the certain routines	22.7%
Interest in rotating objects	20.8%
Turning around	19.3%
Swaying in place	17.3%
Preferring loneliness	15.3%
Inability to make eye contact	13.9%
Inability to play imaginary games	11.4%
Carefree going to strangers	8.9%
Toe walking	6.9%
Sensitivity to high sound	5.9%

In the correlation analysis, there was a positive and significant relationship between paternal age and age at diagnosis ($\rho = .235, p < .001$). However, the relationship between maternal age and the age at diagnosis was not significant ($\rho = .079, p = .264$) (Table V).

Discussion

In the present study, the mean age at diagnosis was 36.76 ± 15.30 months. A review of the literature, including 42 studies that were conducted between 1990 and 2012, reported that the mean age at diagnosis for ASD ranges from 38 to 120 months and that in recent years, ASD has indeed been diagnosed earlier.¹³ A more recent meta-analysis, including studies between 2012 and 2019, also revealed a current mean age at diagnosis of 60.48 (30.90 - 234.57) months.¹⁴ Studies investigating the age at diagnosis of ASD in Türkiye are scarce, although two studies reported a mean age at diagnosis of ASD of 40.70 (12 - 96) months and 32.39 (18 - 48) months, respectively.^{15,16} Based on these findings, it can be said that the mean age at diagnosis has not decreased over time, as expected.

Table IV. Variables affecting age at diagnosis.

	<36 months (n)	>36 months (n)	
Gender			
• Female	22	15	
• Male	95	70	p=0.492
Socioeconomic status			
• Low	62	56	
• Medium/ High	55	29	p<0.05
Mother's education level			
• Middle school and below	77	60	
• High school and above	40	25	p=0.473
Father's education level			
• Middle school and below	47	45	
• High school and above	70	40	p<0.05
First to suspect ASD			
• Mother/ Father	97	47	
• Others	20	38	p<0.001
Parental refusal/denial			
• No	81	44	
• Yes	36	41	p<0.01
History of epilepsy			
• No	108	83	
• Yes	9	2	p=0.087
History of premature birth			
• No	105	74	
• Yes	12	11	p=0.553
History of regression			
• No	84	61	
• Yes	29	18	p=0.648
Has the patient been seen by a pediatrician in the last 6 months?			
• No	23	12	
• Yes	94	73	p=0.304
Has a child psychiatry evaluation been recommended by the pediatrician?			
• No	85	63	
• Yes	32	22	p=0.816
Has the patient been seen by the family physician in the last 6 months?			
• No	32	19	
• Yes	85	66	p=0.402
Has a child psychiatry evaluation been recommended by the family physician?			
• No	93	66	
• Yes	24	19	p=0.752
Chi square test			

Table V. Relationship between age at diagnosis, maternal age and paternal age.

	1	2	3
1.Age of diagnosis (months)	-	-	-
2. Mother's age (years)	0.079	-	-
3. Father's age (years)	0.235*	0.541**	-

Spearman correlation analysis, *p<0.01, **p<0.001

Socio-familial factors

Socioeconomic status and parental education are known to be associated with age at diagnosis of ASD. Most of the families evaluated in the current study had a low socioeconomic status (SED). It was determined that the children of families with middle and higher SED were diagnosed significantly earlier. These results seem to be in line with some studies that found low household income to be a factor delaying the age at diagnosis^{4,17}, in contrast to studies that argue that there is no relationship between household income and age at

diagnosis.^{18,19} It was thought that this result might be related to easier access to health services for families with medium-high SES. The relationship between the early diagnosis of ASD and the educational status of the parents was examined. It was determined that children of fathers who graduated from high school and university, were diagnosed significantly earlier than children of fathers with secondary school and lower education. This result is consistent with the literature.^{20,21} Unlike fathers, there was no significant relationship between the education level of mothers and age at diagnosis. Different results have been reported in the literature regarding the effect of maternal education. In addition to studies that found higher maternal education to be associated with earlier diagnosis²² and maternal education less than college to be associated with later diagnosis²³, there are also studies that did not find a relationship between maternal education and age at diagnosis.²⁴⁻²⁶ The absence of a relationship between maternal education status and age at diagnosis in our study, suggests that mothers may be more sensitive to differences in the development of their children, and this

sensitivity is independent of their education. On the other hand, sensitivity of fathers about the development of their children appears to be related to their education.

In our study, we investigated who initially noticed the signs of autism in their children. It was found that it was mothers in 62.4% of the cases, fathers in 8.9% and others (teachers, health care personnel) in 28.7% of cases. This finding was quite understandable considering that in the majority of the study group, the mother was the caregiver (94%). In a study in our country, it was found that those who first noticed ASD symptoms were mostly mothers (52.7%), and only 1.4% of fathers were the first to notice the symptoms.¹⁶ The age at diagnosis was found to be significantly higher when the parents evaluated the symptoms as normal and the symptoms were noticed for the first time by individuals other than the parents. The failure of parents to notice the symptoms may be related to the fact that families do not have sufficient knowledge and experience about the development of their children. Another reason may be related to the severity of ASD. Symptoms of clinically milder cases may be normalized by families, or associated with non-autistic conditions.

In our study, the relationship between parental age and age at diagnosis was also examined. While a positive relationship was found between paternal age and age at diagnosis, no relationship was found between maternal age and age at diagnosis. In the literature, there is no definite data on the relationship between paternal age and age at diagnosis. In addition to studies claiming that older maternal age is associated with early diagnosis²⁷, there are also studies claiming that there is no relationship

between maternal age and diagnosis.^{21,23} Further studies are needed to demonstrate the relationship between parental age and age at diagnosis.

Medical factors

In our study, 23.3% of the families stated that there was a regression accompanied by the loss of acquired skills in their children. These rates were similar to those found in the literature.^{28,29} While the presence of regression was found to be associated with early diagnosis in the literature^{22,23}, there was no relationship between regression and age at diagnosis in our study. These results are provocative considering that the loss of acquired skills can be a serious concern for families and therefore pushes them to seek remedies more quickly. The most important reason for this situation may be that families perceive the regression period as temporary. Another reason may be related to the fact that families describing regression frequently apply to other departments, especially neurology, aside from the Department of Child Psychiatry, and the detailed neurometabolic examinations are time-consuming. Therefore, if deemed necessary it is suggested that conducting the Child Mental Health examinations, together with these examinations, without leaving them for after the neurometabolic examinations, would help prevent diagnostic delays.

Epilepsy was diagnosed in 5.4% of the cases. In other studies, it was reported that epileptic seizures accompany ASD at a rate of 5 to 30%, and this rate is approximately 20 times higher than the normal population.^{15,30} The most important reason for the low rate of patients diagnosed with epilepsy in our study may be the exclusion of patients with overt neuromotor retardation and diagnosis of cerebral palsy. The age at diagnosis was similar between the group diagnosed with epilepsy and the group not diagnosed and this result is consistent with the literature.²⁷ A history of preterm birth was detected in 11.4% of the cases and these rates are also consistent with the literature, reporting that prematurity is one of the environmental

factors that cause an increase in the incidence of ASD.³¹ Follow-up studies on the relationship between autism and prematurity report that ASD is seen at a rate of 5 to 20% in prematurity, and these rates are 10 to 12 times higher than the prevalence of ASD in the general population.³² In our study, no difference was found between the age at diagnosis between the ASD group with a history of prematurity and the group without. In the presence of a history of prematurity, it's common for families to experience more anxiety about the development of their children and to apply more readily to health centers. Therefore, the age at diagnosis of ASD may be expected to be earlier in this group. Despite this, the lack of difference may be related to the fact that these families' concerns are often about the physical development of their children and their search for remedies for the difference in their emotional development is more limited. The tendency of families to attribute the difference in the cognitive development of their children to prematurity can also be considered as another factor.

In our study, parents' approach to symptoms during the pre-diagnosis period was also questioned. It was found that in 38% of the patients, at least one of the parents denied the symptoms and insisted that their child's development was normal. The age at diagnosis was found to be significantly higher in this group. Similarly, in the literature, parents' concern about social skills development and worry about initial symptoms, are associated with early diagnosis.^{25,33} The reason for this may be that parents cannot see the developmental differences in their children or that they reject the differences in their children, fearing that they will be accused of not raising "good" and "healthy" children. Therefore, persistent denial of the symptoms can be considered as a reflection of defence mechanisms on the part of the parents.

Factors associated with the health care system

Another finding of the study is regarding the examinations of the family physician and

pediatrician in the 6-month period prior to the diagnosis. According to the detailed history and electronic information systems analysis taken from the family, 165 of the children diagnosed with ASD were evaluated by a pediatrician and 151 by a family physician at least once. It was found that 32.7% of the cases evaluated by pediatricians and 32.5% of cases evaluated by family physicians, were referred to a child psychiatry examination. It was determined that some of the families applied for a child psychiatry examination without delay as a result of these referrals, while some of them did not comply with the recommendation. Although our results suggest that the recognition of ASD has increased compared to previous studies, it has been suggested that children with ASD are not yet sufficiently recognized by both physician groups that they frequently encounter. The most important reason for this situation may be that physicians are unable to allocate enough time to patients as a result of the workload they face, often focus on the complaint of the family and cannot find time to make a general evaluation. In fact, it is especially emphasized that the symptoms in children with ASD can only be noticed if sufficient examination times are allocated, otherwise they may be easily overlooked.^{34,35} In addition, the lack of sufficient knowledge and experience on ASD was considered as another factor. In the studies conducted in our country, it was stated that the knowledge and skills of health professionals in recognizing autism spectrum disorder were insufficient.^{36,37} It's reported that only 19% of children diagnosed with ASD had their autism symptoms noted by the pediatricians who examined them, and as a result, it was concluded that pediatricians displayed insufficient effectiveness in recognizing ASD symptoms and directing them.¹⁶ In a study conducted with family physicians, deficiencies were reported in terms of recognizing and correctly guiding ASD.³⁸ As of December 2019, 98.8% of family physicians throughout the country were trained by a team of professionals, including child psychiatrists, within the scope of the nationwide

program for the early recognition, orientation and follow-up of autism.³⁹ Considering that our study was conducted after this training program was completed, it can be said that there still remains a need for training activities for family physicians. In many studies, it has been stated that the concerns expressed by parents about the development of their children may lead to a delay in the diagnosis in cases, where the physicians did not adequately and carefully consider them.^{36,40-42} When the family structures of children diagnosed with ASD were examined, it was found that 17.8% of them were extended families and 82.2% of them were nuclear families. In our study, it was determined that the family's structure didn't affect the age of diagnosis.

Complaints leading to diagnosis

The complaints expressed by the families of the cases diagnosed with ASD at the time of admission to the outpatient clinic were also compiled. It was hypothesized that knowing which symptoms are frequently expressed by families and which symptoms are less frequent, may be a warning for early diagnosis of ASD, especially for healthcare professionals. In our study, when the complaints of the family were evaluated in general, it was seen that they frequently expressed symptoms such as speech delay and hyperactivity, whereas they expressed autism-specific symptoms at a very low rate in most of the cases. Our most common complaint from the families was speech delay and, in a number of studies conducted with groups with ASD, it has been reported that the main reason for bringing the patient to the doctor is the delay in speaking.^{6,26} Therefore, it's imperative to keep a diagnosis of ASD in mind in children with speech delay. The misconception that boys speak late compared to girls and that this is normal among family and some health professionals, causes delays in the diagnosis of ASD in our country. Another complaint frequently expressed by families diagnosed with ASD in our study was hyperactivity. In the

literature, excessive activity (hyperactivity) and its association with ASD have been reported as a very common condition.⁴³ This complaint, which is not specific for the diagnosis of ASD, is one of the most common reasons for presenting to Child Mental Health outpatient clinics. Easily recognizable symptoms such as not reacting to what is being said, not playing imaginary games, not making eye contact, turning around and rocking, were mentioned at a very low rate by our families. Often, these types of symptoms are interpreted by families incorrectly as "hyperactivity" and this may lead healthcare professionals away from the suspicion of ASD and cause delay in the diagnosis. This finding leads us to believe that the complaint of hyperactivity is perceived as an acceptable symptom for large social segments and is more easily expressed.

Our results reveal that the age at diagnosis of ASD is below the target level for early diagnosis, despite numerous studies shedding light on the topic. We have concluded that efforts should be focused on increasing the sensitivity of parents, especially fathers, on the subject in order to reduce the age at diagnosis in our country. There is a need to increase the awareness and experience of ASD-related healthcare professionals, who are likely to encounter children with ASD, and therefore have a critical role in early diagnosis.

Limitations and strengths

Not using a structured assessment tool for the diagnosis of autism, not evaluating the severity of autism symptoms, and collecting some data based on the family's statement are considered to be the most significant limitations.

The size of the sample, the diagnosis with at least two interviews, the participation of a second child psychiatry specialist in the diagnosis process, the use of screening scales during the diagnosis and the evaluation of all patients with a developmental test, are considered the strengths of the study.

Ethical approval

The study was approved by the Clinical Research and Ethics Committee of the Mersin City Training and Research Hospital, Mersin, Türkiye (Date of Approval: March 3, 2021; Reference number/Protocol No: 194). Written consents were obtained after detailed information was given about the subject and purpose of the study.

Author contribution

The authors confirm contribution to the paper as follows: study conception and design: KM, DP, AB, KMZ; data collection: KM, DP; analysis and interpretation of results: DP, AB, KMZ; draft manuscript preparation: KM, DP, AB, KMZ. All authors reviewed the results and approved the final version of the manuscript.

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Conflict of interest

The authors declare that there is no conflict of interest.

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