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EARLY DIAGNOSED AUTISM SPECTRUM DISORDER: THE ROLE OF THE FAMILY PHYSICIAN

ERKEN TANI ALAN OTİZM SPEKTRUM BOZUKLUĞU: AİLE HEKİMİNİN ROLÜ

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SUMMARY

Introduction: *Autism spectrum disorder (ASD) is one of the neurodevelopmental disorders, and the essential features are persistent impairment in social communication and social interaction, and restricted, repetitive patterns of behavior, interests, or activities. To obtain positive results from treatment interventions, early diagnosis of autism spectrum disorder (ASD) is extremely crucial. Diagnosis at early years of childhood can be made with the help of screening tools. However, in Turkey, regular ASD screening does not take place in the scope of primary care services.*

Case: *This case report describes an 11-month-old girl with autism spectrum disorder who was referred from family physician at very early age.*

Conclusion: *It is important that ASD diagnosis, especially for children younger than 3 years of age, is conceived by family physicians and children who are doubted should be urgently guided to a child psychiatrist.*

ÖZ

Giriş: *Otizm spektrum bozukluğu (OSB) nörogelişimsel bozukluklardan biridir. Temel özellikleri sosyal iletişim ve sosyal etkileşimde sürekli bir bozulma ve davranış, ilgi alanı ya da aktivitelerde kısıtlı, tekrarlayıcı kalıplarla gider. Tedavi müdahalelerinden olumlu sonuç almak için OSB'nin erken tanısı çok önemlidir. Tarama testlerinin yardımıyla çocukluk çağının erken dönemlerinde tanı konabilir. Ancak Türkiye'de birinci basamak tedavi hizmetlerinde OSB'nin düzenli taranması yer almamaktadır.*

Olgu: *Aile hekimi tarafından çok erken yaş döneminde yönlendirilen otizm spektrum bozukluğu tanısı alan 11 aylık kız olgu sunulmuştur.*

Sonuç: *Özellikle 3 yaş altında olan çocuklarda aile hekimleri tarafından OSB tanısının akla gelmesi ve şüphe edilen çocukların en kısa sürede çocuk psikiyatristine yönlendirilmeleri önem taşımaktadır.*

INTRODUCTION

Autism spectrum disorder (ASD) is one of the neurodevelopmental disorders, and the essential features are persistent impairment in social communication and social interaction, and restricted, repetitive patterns of behavior, interests, or activities (1). Early intervention has significant benefits for children with ASD such as improving cognitive and adaptive behavior and reducing severity of ASD diagnosis (2). There is a great task for family physicians to examine and observe infant during routine developmental follow-up in terms of autistic symptoms. Therefore, Ministry of Health suggests that following 3-items questioning should be done at least once between 18-36 months to screen ASD in primary health care; looking when his/her name is called, keeping on eye contact and looking at objects pointing at in Turkey (3). The American Academy of Pediatrics Council on Children with Disabilities recommends screening all children for autism at 18 and 24 months using a validated autism screening tool (4). Screening tests are used for early detection of ASD. Checklist for Autism in Toddlers (CHAT) and Modified CHAT (M-CHAT) are applied after 18th months as a screening test for ASD (5,6). However, the most important point in the diagnosis of ASD is being suspected and come to mind. An 11-month-old case who was referred to child psychiatrist by family physician in terms of autistic symptoms in a very early stage is presented.

CASE

A 11-month-old girl was referred for psychiatric assessment to child and adolescent psychiatry outpatient clinic by family physician due to the complaints of not recognizing her mother, not making eye contact.She was born in normal vaginal delivery, 2400 gr weight with asphyxia and cyanosis. Later, she was hospitalized for respiratory failure in neonatal intensive care unit for 22 days. She was treated in paediatric inpatient clinic with the diagnosis of bronchitis when she was 2-month-old and 10-month-old. Once developmental milestones are examined, it has been detected that she could hold her head up at 4 months and sit with support at 6 months. She could neither sit without any support nor crawl. It has also been observed that there was no spelling and no wording; she couldstart gurgling by 8 months, didn't have a social smile, couldn't recognize the mother and respond to her name. Furthermore, she could follow objects and head for sounds but she couldn't reach objects and couldn't hand on them either. During the examination, stereotypic act of swinging was detected. In order to evaluate her cognitive development Ankara Developmental Screening Inventory was used (7). Medium-level of delay has been determined in her language development, psychosocial development, fine-motor and gross-motor skills. According to DSM-5 diagnostic criteria, it corresponds to autism spectrum disorder and delay in cognitive development on medium-level. Paediatric neurology consultation has been requested to search for organic aetiology. Due to detecting hypotonic, dysmorphic face shape and short neck in her neurologic examination, no dysregulation with neurometabolic test battery (TANDEM mass, biotinidase activity) was determined. In her thorough investigation for syndromic baby and hypotonic infant diagnosis, it was observed that female microarray result was normal and result for the cytogenetic was 46,XX. Auditory brainstem responses test was applied in order to evaluate her hearing and both ears were confirmed to be normal. It has been also reported that, although strabismus was detected in her ophthalmologic examination, her vision was well. Her diagnoses were moderate developmental delay, autism spectrum disorder (Level 3) and probable neurometabolic disease. She was referred to special education programme.

DISCUSSION

Recently, studies showed that prevalence of ASD is increasing around the worldwide (8,9). In Turkey, no comprehensive study on prevalence has been conducted. Positive developments seen during the progress of the disease owing to the early diagnosis has shown how important it was to be detected as early as possible (10). In order to achieve this, knowledge levels of the professionals that deal with the children are considered significant and should be increased. According to a research carried out with family

physicians in Kansas City, USA, the reasons behind autism screening were listed as lack of time, inadequate training and lack of funding (11). Availability of time, comfort with screening tool use, previous use and knowledge about specific tools are detected as influencing factors for ASD screening by paediatricians in a study conducted in Canada (12).

Erden et al. found that 58.8% of children with ASD were monitored by paediatricians, and only 4% were referred to a child psychiatrist (13). Child psychiatry rotation is not included in residency training programme of family physician in Republic of Turkey. For this reason, clinical drawbacks about identifying ASD and other childhood psychiatric disorders are existent. However, by arranging education programs after specialty and conducting rotation for child psychiatrists during their specialty education, clinical awareness about these diagnoses can be achieved.

Mental retardation and neurological disorders are often concurrent with ASD (14). Similarly, in our case ASD was accompanied with moderate global developmental delay and neurometabolic disorders, too. Families may not recognize ASD symptoms in their children until they are 2-3 years old, but with the help of observations done by family physician during periodic follow-up visits and questions directed to the family, symptoms that evoke ASD can be detected. In this case which was presented, avoidance of eye contact and not recognizing the mother were two major indications to suspect autism. The reason behind presenting this case is to create awareness on how important it is for family physicians to avoid waiting for 18 months and refer clinically suspected cases to child psychiatrists, independent from screen tests.

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Sorumlu yazar

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