Delayed Referral of Congenital Anomalies

Konjenital Anomalilerde Başvuru Gecikmesi

Y. Hakan ÇAVUŞOĞLU, MD, Assoc.Prof.,[a]
Ayşe KARAMAN, MD, Assoc.Prof.,[a]
İbrahim KARAMAN, MD, Assoc.Prof.,[a]
Derya ERDOĞAN, MD, Msc,[a]
İsmet Faruk ÖZGÜNER, MD, Assoc.Prof.[a]

[a]Department of Pediatric Surgery,
Dr. Sami Ulus Maternity and
Children's Hospital, Ankara

Geliş Tarihi/*Received:* 05.09.2011
Kabul Tarihi/*Accepted:* 30.12.2011

Yazışma Adresi/*Correspondence:*
Y. Hakan ÇAVUŞOĞLU, MD, Assoc.Prof.
Dr. Sami Ulus Maternity and
Children's Hospital,
Department of Pediatric Surgery, Ankara,
TÜRKİYE/TURKEY
hakancavusoglu@hotmail.com

ABSTRACT Objective: Standardized international guidelines recommend a routine physical examination of all newborns before discharge, at least within the first 48 hours of life. The aim of this study was to determine the extent of delayed diagnosis in newborns with early detectable congenital anomalies. **Material and Methods:** Neonates admitted to our pediatric surgery clinic with a diagnosis of congenital anomalies between 2005 and 2010 were retrospectively reviewed. The cohort consisted of patients with imperforate anus, esophageal atresia and congenital abdominal wall defect. A diagnosis made 48 hours or later after birth was considered late diagnosis. **Results:** A total of 236 newborn patients, requiring surgery for congenital anomalies, were admitted to the clinic during this period. A total of 119 patients had anomalies that would be expected to be diagnosed at an early stage. The diagnosis was delayed in 20.2%. In 8 cases, although the diagnosis was early, the baby was taken home by the family. In total, the diagnosis was not made appropriately and on time in 26.9%. The time of diagnosis was 3-15 days in those with a delayed diagnosis (median 4.5 days). Eleven patients with prenatal diagnosis were diagnosed on time. The inpatient duration and survival rates were not different. **Conclusion:** Despite the early diagnosis in patients with a prenatal diagnosis, babies who needed a multidisciplinary approach were born in hospitals without newborn or pediatric surgery departments. Diagnostic delays can be reduced by starting an awareness program among health workers for neonatal surgical problems.

Key Words: Abnormalities; diagnosis; infant, newborn, diseases; esophageal atresia; anus, imperforate

ÖZET Amaç: Standardize uluslararası kılavuzlar, tüm yenidoğanlara, hastaneden taburcu olmadan önce ve her koşulda yaşamın ilk 48 saatinde rutin fizik muayene yapılmasını önermektedir. Çalışmanın amacı, yenidoğanların erken tespit edilebilir konjenital anomalilerinde tanı gecikmesinin boyutunu tespit etmektir. **Gereç ve Yöntemler:** Çocuk cerrahisi kliniğimize 2005 ve 2010 tarihleri arasında konjenital anomali tanısıyla başvuran yenidoğanlar retrospektif olarak değerlendirildi. Bu kohort, imperfore anüs, özofagus atrezisi ve konjenital karın duvarı defekti olan hastalardan oluşmaktadır. Doğumdan sonraki 48. saatte ve daha sonrasında tanı konulması gecikmiş tanı olarak kabul edildi. **Bulgular:** Bu dönemde 236 yenidoğan hasta cerrahi gerektiren konjenital anomali tanısıyla kliniğimize başvurmuştu ve 199 hastada erken dönemde tanı konulması beklenen anomali vardı. Bunların 24 (%20,2)'ünde tanı gecikmişti. Ayrıca sekiz hastada tanı ilk 48 saat içerisinde konulmuş ama hasta ailesi tarafından evden getirilmişti. Toplam olarak hastaların %26,9'unda tanı zamanında ve uygun şekilde konulmamıştı. Gecikmiş tanı konulanlarda tanı konulma süresi 3-15 gündü (ortanca 4,5 gün). On bir hastanın prenatal tanısı olmasına ve hiçbirinde tanı gecikmesi olmamasına rağmen, hastaların %55'i yenidoğan uzmanı, %45'i ise pediatri uzmanı tarafından refere edilmişti. Yatış süresi ve yaşam oranı açısından hastalar arasında fark yoktu. **Sonuç:** Erken tanı konulması gerekli konjenital anomalilerde tanı gecikmesi düşünülenden daha yüksek bulunmuştur. Prenatal tanılı hastalarda tanı gecikmesi olmamasına rağmen, multidisipliner yaklaşım gerektiren bu hastalar, yenidoğan ve çocuk cerrahisi bölümü olmayan hastanelerde doğmuştur. Klasik bilgilerimize aykırı olarak geç tanı konulmuş yenidoğanlarda yatış ve yaşam oranlarında farklılık olmaması, acaba uygun olarak tanı konulmamış yenidoğanlar çocuk cerrahlarına ulaşmadan hayatını kaybediyor mu sorusunu akla getirmektedir. Sağlık çalışanlarına yönelik olarak, yenidoğanda konjenital anomalilere ilişkin farkındalık programları tanı gecikmelerini azaltabilir.

Anahtar Kelimeler: Anormallikler; tanı; bebek, yenidoğan, hastalıklar; özofagus atrezisi; anüs, imperfore

doi: 10.5336/medsci.2011-26349

Turkiye Klinikleri J Med Sci 2012;32(3):745-9

The hospital stay of the mother and her healthy newborn infant should be long enough to allow identification of early problems and to ensure that the family is able and prepared to care for the infant at home.[1] Standardized international guidelines recommend a routine physical examination of all newborns before discharge, at least within the first 48 hours of life.[2] The aim of this study was to determine the extent of delayed diagnosis in newborns with early detectable congenital anomalies.

MATERIAL AND METHODS

Neonates admitted between 2005 and 2010 to the Department of Pediatric Surgery of Dr. Sami Ulus Maternity and Children's Hospital with a diagnosis of congenital anomalies were retrospectively reviewed. The cohort consisted of patients with anorectal malformation (ARM), esophageal atresia (EA) and congenital abdominal wall defect (CAWD). A diagnosis made 48 hours or later after birth was considered late diagnosis. All medical records were evaluated by the first author. Gender, date of birth, mode of delivery, place of birth, birth weight, gestational week, facility referring to our clinic, time our clinic was consulted, duration of hospitalization, ARM classification and whether the patient outcome regarding survival were recorded.

Our clinic is a high volume tertiary care training clinic that accepts patients from Ankara and the surrounding Central Anatolian region. A total of eight pediatric surgery specialists work at the pediatric surgery clinic.

All children under 18 years of age and all pregnant women until the end of puerperium are covered by social security. Women with inadequate financial means are also financially supported so that they can have their prenatal and postnatal follow-ups. The 2008 demographic and health data for Turkey [3] show that 92% of women had received prenatal care from healthcare staff and this was almost always (90%) provided by a physician. The births had taken place in healthcare institutions in 90% and in 97% in the area referring their patients to our hospital, Central Anatolia. The Caesarean section rate was 37%. The first check-up of the babies following birth was by a physician or nurse in 88% and this rate increased to 95% in our region.

The study was approved by the local commission of research assessment (20.04.2011/60556).

Statistical analysis was performed using the Statistical Package for Social Sciences (SPSS) 16.0 software for Windows (SPSS Inc. Chicago, IL, United States). The Shapiro-Wilks' test was used to determine whether the continuous variables were normally distributed or not. The Mann-Whitney U test was used for continuous variables and Fisher's Exact and Chi-Square tests were used for categorical variables where applicable. The Kruskal-Wallis test was used to determine whether the differences between groups for continuous data were statistically significant or not. A p value <0.05 was considered significant.

RESULTS

A total of 451 newborn patients had been admitted to the clinic during this period and 236 patients had congenital anomalies requiring surgery. A total of 119 patients had one or more of ARM, EA or CAWD, that would be expected to be diagnosed at an early stage. All had been born in healthcare facilities. The male/female distribution was 83 (69.7%) to 36 (30.3%). Gestational week data were present for 117 patients; 32 (27.4%) were preterm, 84 (71.8%) were term and 1 (0.9%) was post-term. Evaluation of gestational week by birth weights revealed that 11 (9.5%) out of the 117 patients were small for gestational age, 96 (82.8%) were appropriate for gestational age and 9 (7.8%) were large for gestational age. The vaginal delivery and caesarean section distribution for 119 patients was 51 (42.9%) to 68 (57.1%). The referral to our clinic was by the pediatrician in 55 (46.2%) patients, the neonatologist in 40 (33.6%), and the pediatric surgeon in 12 (10.1%), while 12 (10.1%) patients had been brought directly to our clinic by their families. A prenatal diagnosis was present in 11/119 (9.2%) patients with the case distribution as nine CAWD, one ARM and one EA. While 103 (86.6%) patients were discharged, 16 (13.4%) patients died.

The diagnosis was made after the first 48 hours (i.e. was delayed) in 24 (20.2%) out of 119 patients. In 8 cases, although the diagnosis was made within the first 48 hours, the family took the babies home (seven ARM and one EA). In total, the diagnosis was not made appropriately and on time in 26.9%. The time of diagnosis was 3-15 days in those with a delayed diagnosis (median 4.5 days). There was no difference in terms of the time of diagnosis for various disorders in those with a delayed diagnosis (p=0.668, Kruskal-Wallis) (Table 1). There was a difference between patients with ARM diagnosed on time and where the diagnosis was delayed in terms of anatomical classification (p=0.013, chi-square) (Table 2). The distribution of the 22 CAWD patients diagnosed on time was 6 umbilical cord hernia cases, 4 omphalocele cases and 12 gastroschisis cases. The respective numbers for the 5 patients diagnosed late were 4, 1 and 0 (p=0.055, chi-square). All eleven patients had a prenatal diagnosis (nine CAWD, one ARM and one EA+TEF) and none in this group was diagnosed late; however, 55% was referred by a newborn specialist and 45% by a pediatrician. The inpatient duration and survival rates were not different between those diagnosed late and on time (p>0.05) (Table 3).

TABLE 1: Time to diagnosis of the disorders diagnosed late (days).

	n	Mean ± SD	Median	Range
ARM	7	5.6±2.4	5.0*	3-9
EA + TEF	10	4.4±2.2	3.5*	3-10
CAWD	5	6.8±5.5	3.0*	3-15
ARM +EA +TEF	1	5.0	5.0	
EA + TEF + DA	1	5.0	5.0	
Total	24	5.3±3.1	4.5	3-15

ARM: Anorectal malformation; CAWD: Congenital abdominal wall defect; DA: Duodenal atresia; EA: Esophageal atresia; SD: Standard deviation; TEF: Trachea-esophageal fistula.

TABLE 2: Comparison of anatomical classification of anorectal malformation patients diagnosed on time and late.

	No delay in diagnosis n=44	Delayed diagnosis n=8
Perineal fistula	17	5
Rectourinary fistula	21	0
Rectovestibular fistula	1	2
No fistula	5	1

DISCUSSION

This study only included EA, ARM and abdominal wall defect in the cohort although there were various other congenital anomalies requiring surgery hospitalized in our clinic. The reason for this is an early diagnosis is expected for these three anomalies with a complete physical examination or when guidelines are followed to check such as "the infant has urinated regularly and passed at least one stool spontaneously" and "the infant has completed at least two successful consecutive feedings, with assessment to verify that the infant is able to coordinate sucking, swallowing and breathing while feeding".[1,4] Gastrointestinal obstruction, cardiopulmonary problems, and other problems that may require a longer period of observation by skilled and experienced professionals were excluded from the study.

There are studies defining the time limit for delayed diagnosis as 48 hours and reporting a delay rate of 21-32% while others accept a period of only 24 hours and report the delay rate as 42%.[2,5,6] We used a diagnostic delay criterion of 48 hours in our study as standardized international guidelines recommend a routine physical examination of all newborns before discharge and at least within the first 48 hours of life.[1,2]

Diagnosis, male/female ratio, gestational age, birth weight according to gestational age, caesarean section rate, inpatient duration and survival rates were similar between groups diagnosed on time and delayed.

As in our four cases, umbilical cord hernia may be missed with an imprecise physical examination. The only late referred omphalocele patient was premature, and probably had to be referred after a stabilization period.

Attention is drawn to the necessity for a complete examination of newborns since ancient times. Writing in the second century, *Soranus,* a Greek physician from Ephesus, recommended anal examination for all newborns.[7] *Lorenz Heister* reported

TABLE 3: Comparison of patients diagnosed on time and late.

	No delay in diagnosis n=95	Delayed diagnosis n=24	p
Diagnosis			0.383*
ARM	40/47	7/47	
EA+TEF	29/39	10/39	
CAWD	22/27	5/27	
ARM+EA+TEF	2/3	1/3	
EA+TEF+DA	0/1	1/1	
ARM+EA+TEF+ DA	1/1	0/1	
ARM+DA	1/1	0/1	
Caesarean section rate	56/95	12/24	0.286†
Male/female ratio	68/27	15/9	0.457†
Gestational Age			0.834*
Preterm	25/94	7/23	
Term	68/94	16/23	
Post-term	1/94	0/23	
Birth weight according to gestational age			0.240*
SGA	11/94	0/22	
AGA	76/94	20/22	
LGA	7/94	2/22	
Prenatal diagnosis	11/95	0/24	0.074†
Referred by			0.022*
Pediatrician	47/95	8/24	
Neonatologist	34/95	6/24	
Family	8/95	4/24	
Pediatric surgeon	6/95	6/24	
Hospital stay (days ± SD)	19.7±26.1	22.9±36.2	0.590‡
Survival rate	81/95	22/24	0.329†
ARM	38/40	7/7	
EA+TEF	26/29	9/10	
CAWD	16/22	5/5	
Multiple anomalies	1/4	1/2	

* Chi-Square test; † Fischer's exact test; ‡ Mann-Whitney U test; AGA: Appropriate for gestational age; ARM: Anorectal malformation; CAWD: Congenital abdominal wall defect; DA: Duodenal atresia; EA: Esophageal atresia; LGA: Large for gestational age; TEF: Tracheo-esophageal fistula; SD: Standard deviation; SGA: Small for gestational age.

that an imperforate anus was "not rarely" observed, and warned obstetricians to perform a complete examination of the newborn baby just after the delivery to prevent a delay in the diagnosis.[8] We would expect a smaller rate of delayed diagnosis of congenital anomalies that should be diagnosed early with the warnings given throughout the ages. We believe the delay is due to noncompliance with the guidelines and not to an inadequacy of these guidelines by themselves.

Although babies with a prenatal diagnosis were diagnosed early, those babies who needed a multidisciplinary approach had been born in hospitals without newborn or pediatric surgery departments. Only 55% of prenatally diagnosed babies were born in a hospital with a neonatology specialist.

Although earlier studies have shown that a delayed diagnosis especially for ARM cases increased the complication rate, there was no difference between

the hospitalization period and mortality in the delayed patients in our group.[2] The lack of a difference in inpatient duration or mortality for babies with a delayed diagnosis leads to the question whether some of the newborns who were not diagnosed properly die before reaching pediatric surgeons.

Five of the late referred seven premature patients were referred by a neonatologist. Whether they were referred after a stabilization period could not be documented. This is the limitation of our study.

Diagnostic delays can be reduced by starting awareness programs on neonatal surgical problems among health workers, specially for general practitioners and delivery room personnel.[9] This can be organized as an in-service training program.

REFERENCES

1. American Academy of Pediatrics. Committee on Fetus and Newborn. Hospital stay for healthy term newborns. Pediatrics 2010;125(2): 405-9.
2. Turowski C, Dingemann J, Gillick J. Delayed diagnosis of imperforate anus: an unacceptable morbidity. Pediatr Surg Int 2010;26(11):1083-6.
3. Akadlı Ergöçmen B, Tezcan S, Çağatay P. [Reproductive health]. Türkiye Nüfus ve Sağlık Araştırması 2008. Ankara: Hacettepe Üniversitesi Nüfus Etüdleri Enstitüsü; 2009. p.143-58.
4. Çakmak A, Zeyrek CD, Karazeybek H. [A case of late diagnosis of oesophageal atresia]. Turkiye Klinikleri J Pediatr 2009;18(1):67-9.
5. Lindley RM, Shawis RN, Roberts JP. Delays in the diagnosis of anorectal malformations are common and significantly increase serious early complications. Acta Paediatr 2006;95(3): 364-8.
6. Wilson BE, Etheridge CE, Soundappan SV, Holland AJ. Delayed diagnosis of anorectal malformations: are current guidelines sufficient? J Paediatr Child Health 2010;46(5):268-72.
7. Soranus E. On the care of the newborn. In: Temkin O. Soranus' Gynecology. 1sted. Baltimore, MD: Johns Hopkins University Press; 1956. p.183-205.
8. Yesildag E, Muniz RM, Buyukunal SN. How did the surgeons treat neonates with imperforate anus in the eighteenth century? Pediatr Surg Int 2010;26(12):1149-58.
9. Gupta A, Narasimhan KL. Unusually delayed presentation of esophageal atresia. Pediatr Surg Int 2004;20(11-12):906.