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Clinical Profile of Newborns with Down Syndrome: A Single-center Retrospective Analysis

Author(s)

Hüseyin Şimşek[1], Şefika Aldaş[1], Mustafa Akçalı[1], Berran Biçer[2], Mehtap Durukan Tosun[1]

Affiliation(s)

[1]University of Health Sciences Türkiye, Mersin City Training and Research Hospital, Clinic of Pediatrics, Mersin, Türkiye

[2]University of Health Sciences Türkiye, Mersin City Training and Research Hospital, Clinic of Pediatrics Nurses, Mersin, Türkiye

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Abstract

Down syndrome (DS) is a genetic disorder characterized by intellectual disability and characteristic facial features. This study aimed to evaluate the demographic and clinical characteristics of newborns with a genetically confirmed diagnosis of DS who were admitted to the neonatal intensive care unit (NICU). This retrospective, descriptive study included 28 newborns with genetically confirmed DS who were admitted to the NICU of a tertiary care hospital between February 1, 2017, and April 30, 2025. Demographic and clinical data including sex, gestational age, birth weight, maternal and paternal ages, length of hospital stay, need for mechanical ventilation (MV) presence of congenital heart disease (CHD), hypothyroidism, prematurity, renal and abdominal ultrasonography (USG) findings, white blood cell (WBC) and platelet counts, and 1st- and 5th-minute APGAR scores were retrospectively reviewed. Of the 28 newborns, 53.6% were male and 46.4% were female. Prematurity occurred in 60.7% of cases. The mean gestational age was 35.5±2.9 weeks and the mean birth weight was 2590±619 g. Cesarean section was the delivery method in 78.6% of cases. The mean maternal and paternal ages were 36.5±6.6 and 41.5±6.5 years, respectively, with 71.4% of mothers aged over 35 years. CHD was detected in 96.4% of newborns, hypothyroidism in 53.6%, and pathological renal USG findings in 32.1%. CHD, hypothyroidism, and pathological renal USG findings were detected in 96.4%, 53.6%, and 32.1% of newborns, respectively. MV was required in 71.4% of patients, with a mean duration of 2±8.3 days. The mean hospital stay was 21.5±17.7 days. WBC counts were mostly normal, whereas thrombocytopenia occurred in 53.6% of subjects. Surfactant therapy was administered to one patient, and 50% of patients were diagnosed with sepsis. Two patients died during follow-up. Newborns with DS may present with multiple systemic complications at birth. Early detection of comorbidities, particularly CHD and endocrine disorders, is essential for treatment planning. Understanding neonatal characteristics in these patients may inform long-term management.

Keywords: Down syndrome, hypothyroidism, congenital heart disease, newborn

Correspondence: Hüseyin Şimşek MD, University of Health Sciences Türkiye, Mersin City Training and Research Hospital, Clinic of Pediatrics, Mersin, Türkiye
E-mail: drhuseyinsmsk84@hotmail.com **ORCID:** 0000-0002-3453-6802

Introduction

Down syndrome (DS), the most common chromosomal abnormality, is caused by trisomy of chromosome 21 and is characterized by intellectual disability and congenital anomalies affecting multiple organ systems. It occurs in approximately 1 in 700-800 live births worldwide, with a similar incidence across ethnic and socioeconomic groups[1,2]. Diagnosis is typically made postnatally based on clinical features; however, prenatal screening and diagnostic methods are also available[3,4]. From birth, newborns with DS may face numerous health challenges. Congenital heart disease (CHD), endocrine disorders (particularly congenital hypothyroidism), gastrointestinal anomalies, hematological abnormalities, immunodeficiencies, and hearing and vision problems are common. Many of these infants are born prematurely or with low birth weight, which increases the need for intensive care and complicates clinical management. Respiratory problems, neonatal sepsis, feeding difficulties, and mechanical ventilation (MV) requirements are also frequently encountered. Hospital stays in these patients are often prolonged, depending on the presence of complications. The first months of life constitute a critical period for these infants, and timely interventions can directly affect their quality of life[5-8].

Highlights

- Down syndrome (DS) is associated with a high prevalence of congenital heart disease (96.4%) and hypothyroidism (53.6%) among newborns with DS.
- Prematurity (60.7%) and low birth weight are common, increasing the need for neonatal intensive care unit care.
- Gastrointestinal anomalies, including duodenal atresia and Hirschsprung's disease, were identified.
- Thrombocytopenia was observed in more than half of the patients (53.6%).

Despite advances in medicine, technology, and global assessments, the birth rate of DS neonates has not decreased. Assessing the clinical characteristics of DS births and comparing their prevalence rates in the literature is crucial for both guiding health policies and for developing follow-up protocols. This is feasible: the demographic, clinical, and laboratory characteristics of DS neonates followed within a tertiary healthcare system were retrospectively evaluated. The data obtained contribute to the diagnosis, treatment, and follow-up rates for these patient groups and provide a basis for ongoing care.

Materials and Methods

This retrospective cross-sectional study included 28 patients with genetically confirmed DS who were admitted to the neonatal intensive care unit (NICU) of a tertiary hospital between February 1, 2017, and April 30, 2025. Data for the included patients were obtained from medical records available in the hospital information system. Data on gender, diagnosis, gestational age, birth weight, parental age, need for MV, CHD, hypothyroidism, renal and abdominal ultrasonography (USG) findings, white blood cell (WBC) and platelet levels, 1st- and 5th-minute APGAR scores, and length of hospital stay were recorded retrospectively. Continuous variables were expressed as mean ± standard deviation (SD), and categorical variables as numbers and percentages.

Ethical approval for this study was obtained from the Toros University Scientific Research and Publication Ethics Committee Institutional Review Board (approval number: 2025-05/91, date: 15.05.2025).

Statistical Analysis

Data analysis was performed using SPSS version 25.0 (IBM Corp., Armonk, NY, USA). Continuous variables were expressed as mean ± SD, and categorical variables as frequencies and percentages.

Results

Of the 28 newborns included, 53.6% (n=15) were male and 46.4% (n=13) were female. Seventeen infants (60.7%) were born prematurely. Two patients were hospitalized for suspected gastrointestinal obstruction, two for hyperbilirubinemia, three for malnutrition, four for cyanosis, one for extreme prematurity, and 16 for respiratory distress. The mean gestational age was 35.5±2.9 weeks, and the mean birth weight was 2590±619 g. Twenty-two infants (78.6%) were delivered via cesarean section. None required resuscitation at birth. The mean APGAR scores at 1 and 5 minutes were 7 and 8, respectively. The mean maternal and paternal ages were 36.5±6.6 years and 41.5±6.5 years, respectively. In 71.4% (n=20) of cases, the mothers were older than 35 years. Two mothers had a history of premature rupture of membranes, and one had preeclampsia.

Ten patients (35.7%) had no antenatal diagnosis. Neonates without an antenatal diagnosis were born to mothers who had no or incomplete antenatal follow-up. Patients hospitalized for other diagnoses (respiratory distress, desaturation, or nutritional intolerance) underwent anomaly screening and genetic analysis because physical examination revealed stigmata of DS.

Congenital heart disease was detected in nearly all patients, with only one infant having no cardiac anomaly. The most common malformation was an atrioventricular septal defect (AVSD). Fifteen infants (53.6%) had congenital hypothyroidism. Duodenal atresia was found in four patients (14.21%), one (3.5%) of whom also had an annular pancreas. Hirschsprung's disease was diagnosed in one (3.5%) patient and anal atresia in another patient. Renal USG revealed pathological findings (e.g., hydronephrosis or an ectopic kidney) in nine infants (32.1%); abdominal USG findings were mostly normal. Hearing screening tests were normal in all cases. Regarding respiratory support, six patients (21.4%) were managed with spontaneous breathing, while 20 patients (71.4%) required MV. The mean duration of ventilation was 22.0±8.3 days (mean ± SD). The mean length of hospital stay was 21.5±17.7 days. Laboratory findings showed that WBC counts were mostly within reference ranges, but thrombocytopenia was observed in 53.6% of patients. One infant (3.5%)

received surfactant therapy. Fourteen infants were diagnosed with sepsis. Two (7.1%) patients died during the follow-up period. The demographic characteristics of newborns with DS are presented in **Table 1**, and their clinical characteristics are presented in **Table 2**.

Discussion

In this study, the demographic, clinical, and laboratory characteristics of newborns with DS followed at a tertiary healthcare center were retrospectively analyzed. The findings were compared with the existing literature and interpreted to contribute to clinical practice in our country. DS is the most common chromosomal abnormality among live births, with an incidence generally reported between 1 in 700 and 1 in 1,000. The presence of multiple systemic anomalies in individuals with DS increases neonatal morbidity and mortality[1].

In our study, 59.3% of infants were male, consistent with the literature. Although many studies report an equal gender distribution, some have indicated a slight male predominance[9].

The mean maternal age in our study was high. This finding supports the association between advanced maternal age and DS. Especially in pregnancies over the age of 35, the risk increases significantly. Similar results have been reported in many studies conducted worldwide and in Türkiye[10-14]. Therefore, more effective implementation of prenatal diagnostic approaches in pregnancies of advanced maternal age is of great importance.

Although the mean APGAR scores were between 7 and 8, some cases had lower scores, which may be associated with cardiac or respiratory problems during the neonatal period.

Table 1.
Demographic characteristics of patients diagnosed with Down syndrome

	(n=28)
Gestational age, weeks*	35.5±2.9
Birth weight, grams**	25.90±619
Gender, M/F (%)	15 (53.6%)/13 (46.4)
Mode of delivery, C/S/n (%)	22 (78.6%)/6 (21.4)
Patients without prenatal diagnosis, n (%)	10 (35.7%)
5-minute APGAR	8
Mother's age (year)	36.5±6.6
Father's age (year)	41.5±6.5
Prematurity, n (%)	17 (60.7%)

*Values are given as mean ± SD. **Values are given as median (minimum-maximum)
C/S: Cesarean section, M/F: Male/female, SD: Standard deviation

Table 2.
Clinical characteristics of patients diagnosed with Down syndrome

	(n=28)
Congenital heart disease, n (%)	27 (96.4%)
Congenital hypothyroidism, n (%)	15 (53.6%)
Duodenal atresia	4 (14.2%)
Hirschsprung's disease	1 (3.5%)
Anal atresia	1 (3.5%)
Renal pathology	9 (32.1%)
Thrombocytopenia	15 (53.6%)
Mortality	2 (7.1%)

Prematurity was observed in 33.3% of newborns with DS. Reported rates of prematurity in the literature vary, with some studies indicating rates between 15% and 30%[15-17]. Prematurity is an important factor that increases morbidity among DS infants and should be considered in follow-up and treatment protocols.

The mean birth weight in our study was approximately 2,750 grams. Although intrauterine growth restriction is not commonly seen in DS, some studies have reported higher rates of low birth weight[17]. This may be related to accompanying cardiac or gastrointestinal anomalies.

The mean length of hospital stay was 14 days. A prolonged hospital stay is generally associated with congenital anomalies, feeding difficulties, or infections. In DS infants, the presence of CHD significantly increases both the need for intensive care and the length of hospitalization[18].

Approximately 65% of neonatal DS cases are admitted to the intensive care unit in the early postnatal period; the most common reasons for admission are CHD, prematurity, and respiratory distress. The length of stay in neonatal intensive care is prolonged depending on the presence of complications and the use of MV, and mortality rates may be up to eight times higher[6].

In our study, the most common accompanying anomaly was CHD, present in 96.4% (n=27) of cases, whereas the literature reports a prevalence of 40-60%. The high rate observed in our study may be attributable to the fact that all hospitalized patients underwent echocardiographic evaluation during their stay. and the reason for our high rate of CHD in children born with DS and undergoing echocardiographic evaluation, patients are already admitted to intensive care due to respiratory problems and heart problems, and many have systemic diseases. The most common types of CHD were AVSD, ventricular septal defect, and atrial septal defect[18,19]. Since cardiac anomalies are determinants of quality of life and prognosis, echocardiographic evaluation is recommended for all newborns diagnosed with DS during the early postnatal period.

The rate of hypothyroidism in our study was 53.6%. Thyroid dysfunction is common in individuals with DS, and congenital hypothyroidism is a critical risk factor for developmental delay[20,21]. Therefore, in addition to national screening programs, repeated evaluation of thyroid function in infants with DS is important.

In our study, duodenal atresia was detected in four newborns with DS; in one of these cases, duodenal atresia was accompanied by annular pancreas. In addition, Hirschsprung's disease was found in one case and anal atresia in another. These findings indicate that the frequency and variety of gastrointestinal anomalies in DS infants should not be underestimated.

Duodenal atresia is reported to be the most common gastrointestinal malformation in DS patients, with a prevalence significantly higher than that in the general population. According to a systematic review, approximately 3% of DS infants have duodenal atresia, and in some regions, such as the Middle East, this rate can be as high as 6%[22]. Another study reported a prevalence of 3.9% among DS cases, with an annular pancreas accompanying approximately 12% of these cases[18,23]. Embryologically, duodenal atresia results from failure of recanalization of the duodenal lumen during the eighth week of fetal life[24]. The presence of annular pancreas in our patient represents a rare but documented association reported in case series of patients with DS.

Hirschsprung's disease occurs at higher rates in newborns with DS. While the rate is 1:5000 in the general population, it can be as high as 1:150 in infants with DS. Failure of neural crest cell migration during embryogenesis is the principal mechanism underlying its association with DS.

Anal atresia is a rare anomaly, but it has been reported among gastrointestinal anomalies in infants with DS, with a prevalence of approximately 1% in the DS population[25].

Based on these findings, systematic prenatal and postnatal evaluations for gastrointestinal anomalies in newborns with DS are critical[25]. In cases of suspected duodenal atresia, the "double bubble" sign on radiological imaging or prenatal ultrasound findings should be promptly evaluated. Furthermore, screening programs should include assessments for Hirschsprung's disease and anal atresia to allow appropriate surgical preparation. The presence of these anomalies highlights the need for a multidisciplinary approach beginning at birth for newborns with DS. Systematic screenings during the prenatal and postnatal periods are crucial for early diagnosis and appropriate surgical planning.

On renal USG, major anomalies were infrequent, although minor findings such as pelvicaliectasis were detected in some cases. Renal malformations associated with DS are rare, but developmental abnormalities of the kidney can lead to serious clinical consequences in some individuals[26].

When laboratory findings were evaluated, WBC counts were mostly within reference ranges, while thrombocytopenia was observed in 53.6% of cases. Hematological abnormalities are common in newborns with DS, and thrombocytopenia, in particular, requires careful monitoring due to the risk of transient myeloproliferative disease and leukemia[27,28].

All hematologic abnormalities in our patients returned to normal within the second week of hospitalization. Therefore, none of our patients were considered to have myeloproliferative disease.

In our study, 2 of the 28 newborns with DS (7.1%) died during the neonatal period. Published studies indicate that mortality among newborns with DS is associated with additional risk factors, including prematurity, severe CHD, gastrointestinal malformations, immune deficiencies, and infections[13,25]. Especially among infants who require intensive care during the first weeks of life, complications such as MV and sepsis have been reported to increase mortality[29].

The reason why the prematurity, CHD incidence and anomaly rate in the patients we follow are higher than in the literatüre, since our hospital is a city hospital with regional hospital status and a perinatal center, the follow-up of risky pregnancies and the births of patients who may require intensive care are accepted to our hospital by referral.

In our series, one of the deceased cases had severe CHD, while the other was premature and had severe respiratory failure. These findings are consistent with the risk factors reported in the literature. Furthermore, these cases demonstrate that newborns with DS are not solely affected by a chromosomal abnormality but also require systemic evaluation and multidisciplinary follow-up, both of which are vitally important. Early intervention, rapid diagnosis, and supportive treatments during the neonatal period are considered decisive in improving survival in this group.

Study Limitations

One of the most significant limitations of this study is that the data were obtained retrospectively from the hospital information system. The sample size was small because this was a single-center, tertiary-care study that included only patients admitted to the NICU. However, we believe that our patients with conditions severe enough to require intensive care provide important data on DS and associated morbidity. Therefore, we present important data on mortality and morbidity among critically ill patients with DS. Although our data are limited in number, they will contribute to the literature because of their comprehensive coverage of critically ill patients.

Conclusion

Neonates with DS require close monitoring after birth due to their multisystem involvement. Information on CHD, prematurity, use of respiratory support devices, feeding intolerances, structural intestinal pathologies, hematological problems, and clinical assessments may be obtained. Careful assessment and appropriate referral during the neonatal period are critical to improving the survival and quality of life of this infant.

Ethics

Ethics Committee Approval: Ethical approval for this study was obtained from the Toros University Scientific Research and Publication Ethics Committee Institutional Review Board (approval number: 2025-05/91, date: 15.05.2025).

Informed Consent: Because the study was designed retrospectively no written informed consent form was obtained from the patients.

Footnotes

Author Contributions: Şimşek H: Surgical and Medical Practices, Concept, Data Collection or Processing, Analysis or Interpretation, Writing; Aldaş Ş: Surgical and Medical Practices, Concept, Design, Analysis or Interpretation, Literature Search, Writing; Akçalı M: Surgical and Medical Practices, Concept; Biçer B: Concept, Analysis or Interpretation; Durukan Tosun M: Design, Data Collection or Processing.

Conflict of Interest: The authors declare no conflicts of interest.

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