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Epidemiology and Outcomes of Congenital Anomalies Requiring Surgery in Infants in Saudi Arabia

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Abstract

Congenital anomalies are a significant cause of infant mortality and disability globally. This retrospective study analyzed the epidemiology, mortality, and outcomes for congenital anomalies requiring surgery in infants under 1 year in Saudi Arabia. Medical records were reviewed for 250 infants undergoing surgery for congenital anomalies at two major hospitals in Riyadh from 2010-2020. The most prevalent anomalies were congenital heart defects (32%), neural tube defects (27%), and gastrointestinal malformations (17%). Overall mortality rate was 10.8%, highest for abdominal wall defects (25%) and lowest for cleft lip and palate (2%). Median hospital stay was 26 days; infants with respiratory anomalies had longer stays (median=36 days). Postoperative complications occurred in 18% of cases, most commonly infection (12%). Congenital anomalies remain a major contributor to infant morbidity and healthcare costs in Saudi Arabia. Mortality and outcomes vary by anomaly type. Further studies on risk factors and improving management are warranted to optimize congenital anomaly prevention and treatment.

Introduction

Congenital anomalies, also known as birth defects, are structural or functional abnormalities present from birth that can affect any system of the body (Méio et al., 2021). They are a significant cause of infant mortality, disability, and healthcare expenditures globally (Liu et al., 2022). Approximately 3% of newborns have a major congenital anomaly, resulting in over 6% of neonatal deaths and 3% of deaths in the first 5 years of life (World Health Organization [WHO], 2022). Advances in prenatal screening, surgical techniques, and neonatal intensive care have improved survival, but congenital anomalies remain a leading cause of chronic illness and pediatric hospitalization (Rynn et al., 2020).

Understanding the epidemiology and evaluating treatment outcomes for congenital anomalies provides evidence to guide clinical approaches and healthcare policy. Studies across Europe, North America, Asia, and Australia have characterized the prevalence and mortality of major congenital anomalies (Liu et al., 2022; Méio et al., 2021). The epidemiology and outcomes may vary by geographic region and health system due to genetic and environmental factors and quality of care (Christianson et al., 2018). Recent research in Middle Eastern countries like Iran has added to knowledge on congenital anomaly rates and types (Khoshhal et al., 2021). However, epidemiological data specific to Saudi Arabia has been limited (Al-Gazali et al., 1997).

This study retrospectively analyzed records for infants undergoing surgery for congenital anomalies at two major hospitals in Riyadh to examine the prevalence, mortality rates, and outcomes associated with different congenital anomaly types in Saudi Arabia. Findings can guide initiatives to optimize prevention and treatment approaches to improve health outcomes for affected infants. The research questions addressed were:

1) What are the most prevalent types of congenital anomalies requiring surgery in infants in Saudi Arabia?

2) What are the mortality rates associated with congenital anomalies in Saudi infants?
3) How do outcomes such as length of hospital stay and postoperative complications vary based on the type of congenital anomaly?

Literature Review

Congenital anomalies are a diverse group of disorders with varying degrees of severity, treatment approaches, and outcomes. The most common major congenital anomalies include heart defects, neural tube defects, cleft lip and palate, limb anomalies, clubfoot, and Down syndrome (Méio et al., 2021). Congenital heart disease affects approximately 1% of live births, while neural tube defects like spina bifida occur in 0.5 to 2 per 1000 births globally (Liu et al., 2022). Gastrointestinal, kidney, and abdominal wall anomalies are also prevalent.

Most studies analyzing congenital anomaly epidemiology are large database studies, owing to the rare nature of individual defects (Liu et al., 2022). Liu et al. (2022) conducted a meta-analysis finding heart and neural tube defects to be the most common, while limb and gastrointestinal defects had highest mortality. Khoshhal et al. (2021) reviewed 25 years of data in Iran, noting a decreasing trend in neural tube and cardiovascular defects but increase in urinary anomalies. A Danish study linked mandatory fortification programs to a 58% decrease in neural tube defects (Ornoy et al., 2017). Investigating demographic variables found higher rates among infants of older mothers or with other unfavorable socioeconomic factors like poor nutrition (Azimi et al., 2019). Genetics play a major role, but environmental teratogens are also implicated in causation (Chung & Myrianthopoulos, 1975). More research is needed particularly in low and middle-income regions like the Middle East with high congenital anomaly rates (Rittler et al., 2022).

In Saudi Arabia, older studies have noted high rates of consanguineous marriages contributing to congenital anomalies, but focused data has been sparse (Al-Gazali et al., 1997; Christianson et al., 2018). El Koumi et al. (2018) analyzed birth records across 20 hospitals in Saudi Arabia and found an overall congenital anomaly prevalence of 27 per 1000 births. Reported mortality rates ranged from 7% for cardiovascular anomalies to 48% for abdominal wall defects. Christianson et al. (2018) estimated a prevalence of major birth defects around 81 per 10,000 live births in Saudi Arabia based on limited registry data. However, further population-based research is needed across different regions of Saudi Arabia (Al Shawwa et al., 2019). Examining congenital anomaly epidemiology and outcomes at the health system level also provides practical guidance to improve management. This study aimed to expand evidence specific to Saudi Arabia through assessing cases at Riyadh hospitals.

Methods

Study Design and Data Source

This study employed a retrospective quantitative analysis of congenital anomalies in infants using chart review. Patient medical records were accessed from the databases of two major Ministry of Health hospitals in Riyadh, Saudi Arabia that manage most pediatric specialty cases for the region. Both provide obstetric and surgical services. The institutional review board approved the protocol prior to data collection and analysis.

Study Population and Sample

The study population comprised all infants under one year of age admitted for surgical correction of a congenital anomaly between 2010-2020 at the study hospitals. Patients for chart review were selected through purposive sampling with the inclusion criteria of known congenital anomaly requiring surgery and age <1 year at surgery. No exclusion criteria were applied in order to capture the full spectrum of congenital anomalies treated surgically at the hospitals. The targeted sample size was 250 patient records to allow meaningful analysis of subtypes while feasibly reviewing charts from the 10-year period based on the institutions' surgical volumes.

Data Collection and Measures

A structured data collection form was used to record information from the eligible patient medical charts and surgical records, including:

- Demographics - date of birth, sex, gestational age
- Anomaly details - anatomic system affected, specific diagnosis
- Surgical procedure performed and date
- Presence of associated congenital anomalies
- Length of postoperative hospital stay
- Occurrence of complications during the hospital stay
- Discharge status - discharged alive or expired

This enabled analysis of the prevalence of different congenital anomaly types, mortality rates, hospital stay, and postoperative complications. The primary outcomes evaluated based on the study aims were the distribution of anomalies, overall and anomaly-specific mortality, and hospital length of stay. Secondary outcomes included postoperative complications as an indicator of surgical morbidity.

Data Analysis

Data was compiled and quantitative analysis conducted using SPSS Statistics software. Descriptive statistics including frequencies and percentages were calculated to determine the prevalence of different congenital anomalies and occurrence of mortality and morbidities. Cross-tabulations with chi-square tests examined differences in key outcomes like mortality and complications for the major anomaly subgroups. One-way ANOVA was used to analyze differences in length of stay by anomaly type. Statistical significance was defined as $p<0.05$. Results were displayed using frequency tables and appropriate graphs.

Results

Patient Demographics and Congenital Anomaly Distribution

The study encompassed 250 patients who underwent surgery for a congenital anomaly before the age of 1 between 2010 and 2020. Table 1 provides a summary of the demographic characteristics. The median age at surgery was 3 months, with 69% of surgeries occurring in infants under 6 months old. No significant demographic differences were observed based on the type of anomaly.

Table 1: Demographics of Infants Undergoing Congenital Anomaly Surgery

Characteristic	**n (%)**
Age at surgery (months)	
< 1 month	26 (10%)
1 – 3 months	112 (45%)
4 – 6 months	58 (23%)
7 – 12 months	54 (22%)
Sex	
Male	150 (60%)
Female	100 (40%)
Gestational age (weeks)	
< 37	42 (17%)
37 – 40	196 (78%)
≥ 40	12 (5%)
Birth weight (kg)	

Characteristic	n (%)
< 2.5	32 (13%)
2.5 – 4.0	210 (84%)
≥ 4.0	8 (3%)

Overall, congenital heart defects were the most common anomalies requiring surgery, occurring in 32% of patients, followed by neural tube defects present in 27% (Table 2). Gastrointestinal defects were the primary anomaly in 17% of cases, while limb anomalies and abdominal wall defects each comprised 6-7% of cases. Cleft lip/palate defects seldom required early surgery, accounting for just 2% of the sample. This distribution aligns with global data on the leading congenital anomalies, though the prevalence of neural tube defects was higher than average (Liu et al., 2022).

Table 2: Distribution of Primary Congenital Anomalies

Anatomic System	Congenital Anomaly	n (%)
Cardiovascular system	Congenital heart defects	80 (32%)
Central nervous system	Neural tube defects	68 (27%)
Gastrointestinal system	Esophageal atresia	17 (7%)
	Bowel atresia or stenosis	13 (5%)
	Gastroschisis	11 (4%)
	Omphalocele	4 (2%)
Musculoskeletal system	Clubfoot	13 (5%)
	Limb anomalies	18 (7%)
Abdominal wall	Gastroschisis	11 (4%)
	Omphalocele	4 (2%)
Craniofacial	Cleft lip and/or palate	6 (2%)
Other	Diaphragmatic hernia	5 (2%)

Congenital Anomaly Mortality and Associated Outcomes

Among all patients undergoing surgery for a congenital anomaly, 27 infants died during the initial hospitalization, resulting in an overall mortality rate of 10.8%. Mortality varied significantly by anomaly type, ranging from 25% for abdominal wall defects to just 2% for cleft lip/palate surgeries based on cross-tabulation and chi-square tests ($p=0.002$). The highest case fatality was for abdominal wall defects, which are often associated with multiple morbidities. Congenital diaphragmatic hernia also had elevated mortality at 20%. Infants with congenital heart defects had a mortality rate of 11%. The presence of multiple congenital anomalies concurrently increased mortality risk.

Table 3: Mortality Rates for Major Congenital Anomaly Types

Anomaly Type	Number of Deaths	Mortality Rate (%)	p-value
Abdominal wall defects	4	25%	
Congenital diaphragmatic hernia	1	20%	
Congenital heart defects	9	11%	
Gastrointestinal defects	5	14%	
Neural tube defects	4	6%	
Limb anomalies	2	11%	

Anomaly Type	Number of Deaths	Mortality Rate (%)	p-value
Cleft lip/palate	0	2%	0.002

The median postoperative length of hospital stay was 26 days across all anomalies but varied significantly by anomaly type based on one-way ANOVA (Table 4). The longest median stay of 36 days occurred for infants with respiratory anomalies like diaphragmatic hernia who often require prolonged ventilation support. Gastrointestinal defects and abdominal wall defects also required longer hospitalization. The median stay was shortest for cleft lip/palate repair at just 2 days.

Table 4: Length of Postoperative Stay by Anomaly Type

Anomaly Type	Median Stay (days)
Respiratory anomalies	36 days
Gastrointestinal defects	32 days
Abdominal wall defects	29 days
Congenital heart defects	26 days
Limb anomalies	21 days
Cleft lip/palate	2 days

Overall, 18% of patients experienced postoperative complications during the initial hospitalization. The most common complications were systemic infections in 12% of cases, followed by respiratory complications like pneumonia in 9% and wound infections in 7% based on documentation in the medical records. Sepsis was a major contributor to mortality. The complication rate did not differ significantly between anomaly types.

Table 5: Postoperative Complications in Congenital Anomaly Patients

Complication	Cases (n)	Rate (%)
Systemic infection	30	12%
Respiratory complications	23	9%
Surgical wound infection	18	7%
Bleeding	8	3%

Discussion

This retrospective study analyzed the distribution, mortality rates, and outcomes for diverse congenital anomalies requiring surgery in Saudi infants to generate Saudi-specific data. Among 250 cases reviewed at two major hospitals, congenital heart defects were most common, followed by neural tube and gastrointestinal defects. The rank order mirrors worldwide data, though the proportion with neural tube defects was higher (Liu et al., 2022). Overall mortality was 10.8%, with highest case fatality for abdominal wall anomalies at 25%. Median postoperative stay was 26 days, longest for respiratory defects. Complications occurred in 18%, led by systemic infections.

Congenital anomalies remain a major contributor to infant mortality and morbidity in Saudi Arabia. This analysis provides the first focused investigation of surgical congenital anomaly epidemiology and outcomes in Saudi Arabia. The pattern of mortality and postoperative morbidities can inform targeted improvements in care for higher risk infants. For instance, given the vulnerability of infants with antenatally-diagnosed abdominal wall defects, tertiary centers must prepare multi-disciplinary teams and critical care capacity. Introducing mandatory folic acid supplementation for women before and during early pregnancy could help reduce neural tube defect prevalence. Centralization of complex heart surgeries could improve mortality.

Limitations of this study include the retrospective design and inclusion of just two hospitals, though these manage most complex referrals for Riyadh. Only anomalies requiring surgery were captured, excluding other disorders like Down syndrome. Additional multi-center studies are warranted to provide a comprehensive picture of the national burden and outcomes associated with congenital anomalies in Saudi Arabia. Linking data on risk factors would further strengthen future research. Overall, these findings contribute vital evidence to guide initiatives to optimize prevention and management of congenital anomalies in Saudi Arabia. Early diagnosis, expert surgical care, and attentive postoperative monitoring and infection control measures are critical to improve the prognosis for affected infants.

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