Original Article

Prenatal diagnosis of congenital anomalies and their outcome in Tribhuvan University Teaching Hospital

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Abstract

Introduction: Congenital anomalies represent defects in morphogenesis during early fetal life. The proportion of perinatal deaths due to congenital anomalies is increasing in developing countries like Nepal as a result of reduction of mortality due to other causes.

Methods: A retrospective study was conducted in the Pediatric Surgery Unit, Department of Surgery, Tribhuvan University Teaching Hospital (TUTH). Data were collected from patients' hospital files from 2013 to 2015 AD.

Result: A total of 33 patients who had congenital anomalies either diagnosed and referred from outside or diagnosed at TUTH were included. Various types of anomalies involving different body systems were identified during the prenatal period. But surgery was performed in only seven cases.

Conclusion: Due to widespread availability of ultrasound the number of patients with congenital anomalies who are being detected is on the rise. The proper management of these children in the intrauterine and neonatal period is a challenge in our context. Lack of proper policy and resources is a big hindrance to the proper management of these group of patients.

Keywords: Congenital anomalies, Surgery, Nepal

Introduction

Congenital anomalies can be defined as structural or functional anomalies, including metabolic disorders, which are present at the time of birth. Congenital anomalies may be genetic, infectious or environmental in origin and result in long-term disability. Congenital anomalies affect 1 in 33 infants per year[1]. There are 3.2 million birth defect-related disabilities every year[2]. While geographic variations for individual anomalies exist, the global prevalence rates at birth of congenital anomalies are remarkably similar worldwide[3]. According to the Disease Control Priorities (Surgery), 50% of congenital anomalies can be treated with surgery[1].

Many children with congenital anomalies that were once thought incompatible with life are now living normally. Severely injured children are restored to normal function, failed organs are replaced, neonatal surgery has become routine, and the efficacy of fetal surgery is being investigated in clinical trials. Nevertheless, there are still major gaps in the surgical care of children living in developing countries. Pediatric surgery has often been viewed as too expensive and as a non-essential service, and it has been excluded from most child health programmes in such countries[4].

Methodology

A retrospective study was conducted in the Paediatric Surgery Unit, Department of Surgery, Tribhuvan University Teaching Hospital. Data were collected from patients' hospital files from 2013 to 2015 AD.

Results

Thirty three cases either diagnosed and referred from outside or diagnosed at TUTH were included. Antenatal ultrasonography was the diagnostic tool for all the cases. Thirteen cases were of genitourinary anomalies, the most common being hydronephrosis. Thirteen cases were of gastrointestinal anomalies, small bowel atresia being the most common. Four cases with respiratory system anomalies were also included (Table 1). One patient with tracheo- oesophageal fistula was refused for surgery. A patient with congenital diaphragmatic hernia died on the day of delivery. Six patients were managed conservatively (Table 2). Two cases were terminated due to severe anomalies. Fourteen pregnant ladies lost to follow up (Table 3). Seven cases were operated (Table 4). Operated cases were of jejunal atresia, oesophageal atresia, congenital diaphragmatic hernia, clumped bowel mass and high anorectal malformation. Four cases expired post operatively. Two cases of duodenal atresia and one case of high anorectal malformation survived post operatively.

Table 1: Anomalies involving different systems

System Involved	Number of Cases
1 Genitourinary Malformations	13
Unilateral hydronephrosis	5
Bilateral hydronephrosis	3
PUV	3
Multicystic dysplastic kidney	1
Renal mass	1
2 Gastrointestinal Anomalies	13
Oesophageal Atresia	1
Duodenal Atresia	6
Intestinal atresia	4
Gastroschisis	2
Intestinal cyst	1
3 Respiratory System	4
Congenital Diaphragmatic Hernia	3
Tracheo- oesophageal fistula	1
4 Others	2
Complex cystic right iliac fossa mass	1
Mixed anorectal malformation + Multicystic kidney disease	1

Table 2: Conservative Management

Antenatal USG	Findings after Birth
Hydronephrosis	Radiology after birth showed left hydronephrosis most likely moderate to severe with PUJ stenosis
Cystic mass in RIF	Repeat USG: mass persistent not increasing in size
Intestinal Thickening	Normal clinically at birth • 1 month follow up USG normal
B/L hydronephrosis	Lost to follow up
Abdominal cyst	Follow up USG at 1 month normal
Renal mass	Serial abdominal scans described non visualisation of the mass

Table 3: Cases lost to follow up

Case	Period of Gestation at Diagnosis
Gastroschisis	28
Intestinal atresia	36
Abdominal cyst	37
B/L hydronephrosis	37
Right gross hydronephrosis	22
Right gross hydronephrosis	26
Left calyectasis	25
B/L gross hydronephrosis	37
Congenital Diaphragmatic Hernia	23
Duodenal atresia	33
Duodenal atresia	18
Duodenal atresia	24
Gastroschisis	33
PUV	29

Table 4: Operated Cases and their outcome

Operated Cases	
Distal Jejunal Atresia	Operation on day 1 Per op findings: Distal jejunal atresia Patient expired at 6 month
Oesophageal Atresia	Thoracotomy with proximal oesophagostomy with distal ligation with gastrostomy Per op findings: oesophageal atresia Patient expired on post operative day 2
Congenital Diaphragmatic Hernia	Laparotomy with closure of diaphragm with prolene mesh and abdomen closed over silo bag Per op findings: No left hemi diaphragm, liver with bowel loops in left thorax, small abdominal cavity Patient expired on the day of operation
Clumped Bowel Mass	Laparotomy with resection of clumped bowel mass with ileostomy Patient expired on the day of surgery
High anorectal malformation	Sigmoid colostomy Per operative findings: Dilated Sigmoid Colon Patient doing well on follow up with colostomy reversal done
Duodenal Atresia	Operated on day 2 ,Duodenodudenostomy, Patient survived and on follow up
Duodenal Atresia	Operated on day 5 of life,Duodenodudenostomy, Patient survived and on follow up

Discussion

Nepal is one of the poorest countries in the world. According to UNDP's Human Development Index (HDI) 2013, 44% of the population is currently under the poverty line. The per capita income is only 377 dollars. Nepal has made a huge leap in a short span of time in decreasing infant mortality rate and has already attained the millennium development goal. The history of paediatric surgery in the country is quite short. There are only a few centres in the country which offer paediatric surgery facilities. The number of centres which offer neonatal surgeries are even less.

According to a study by Loefler health care policy in developing countries does not reflect the surgical needs of children and there has been a general failure to recognize the importance of surgical care in the developing world, where it tends to be viewed as a luxury. It is often forgotten that surgery is an essential component of basic health care[5]. There is a chronic shortage of qualified paediatric surgeons in many developing countries[6]. In the Global Burden of Disease (GBD) study, childhood surgical conditions were underrepresented[7]. A study by Basnet et al showed that 1.3% of babies delivered at TUTH had congenital malformation responsible for about 15% of the perinatal mortality[8]. A commonly held but erroneous view is that congenital disorders are not a public health issue in developing countries. A study by Penchaszadeh et al. showed an epidemiological transition with significant declines in infant mortality rates due to reduction of infections and malnutrition and a relative increase of morbidity and mortality due to non-communicable diseases, including congenital anomalies[9]. Numerous experiences indicate that the conditions of underdevelopment require preventive modalities of low cost and high impact, which may differ from those implemented in wealthier countries[10]. Rather than training paediatric surgeons, the solution for developing countries may be to re-establish general surgery to deal with the operative management of common surgical conditions[11]. Paediatric surgical education can be strengthened through the work of donor organizations[12]. A group convened by the World Health Organization (WHO, 1999) stated that programs for the prevention of birth defects should be implemented when infant mortality falls below 40 per thousand,

essentially because it is at that stage that congenital anomalies begin to make up a significant proportion of infant mortality[13].

Though Nepal has made significant progress in preventing congenital anomalies through programs such as folic acid supplementation during pregnancy other varied causes need to be addressed. Besides very little has been done to manage these conditions once they are detected either in the antenatal period or after delivery.

Conclusion

Due to widespread availability of ultrasound the number of patients with congenital anomalies who are being detected in a developing country like Nepal is on the rise. Experiences in developed countries have shown that if cases are timely managed in ante natal and post natal period success can be achieved. Besides for proper management these conditions should be recognized as a significant problem in the health policy of our country. An effective low cost and high impact program should be initiated. There should be appropriately trained surgeons for dealing with these cases with special requirements. Cases with malformations involving multiple systems which are not amenable to surgery should be terminated. Just as the World Bank has defined a package of basic health care, an essential package of pediatric surgical services for developing countries such as should be defined. Both preventive and curative services are required.

The details of how surgical care should be integrated into child health programmes, and its exact role remain to be determined. Whatever its role, however, it should be evidence-based, cost-effective and work towards benefiting the largest possible number of children.

Conflict of interest: None declared.

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