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Original Article

Associated Anomalies and Clinical Outcomes in Infants with Omphalocele: A Single-centre 10-year Review

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Abstract

Objective: To examine the perinatal characteristics, associated anomalies and clinical outcomes of newborns with omphalocele admitted to Queen Mary Hospital (QMH), Hong Kong during a 10-year period. ***Methods:*** We identified all newborns with omphalocele who were admitted to the neonatal intensive care unit at QMH from 2005 to 2014. Maternal and patient demographic data, associated anomalies and outcome data were reviewed retrospectively. ***Results:*** A total of 19 infants with omphalocele were identified. Median gestational age at birth was 38 weeks with a median birth weight of 3140 g. Fifty-three percent (10/19) were diagnosed with at least 1 other anomaly, with congenital heart defect being the most common associated anomaly. Median age at first operation (either primary closure or application of silo) was on day 1 of life, with delayed closure in staged operations being carried out at a median age of 8 days. Infants with non-liver containing omphaloceles were more likely to have primary repair compared to those with liver-containing ones (7/8 [88%] vs. 4/11 [36%], p=0.03). Overall survival rate at discharge was 84% (16/19). Two out of 3 cases died of lethal congenital anomaly (alveolar capillary dysplasia) while the other one suffered from postoperative midgut volvulus. Postoperative complications occurred in 6 patients (6/17 [35%]), with ventral hernia being the most common complication. Long-term medical problems including failure to thrive (6/12 [50%]), gastroesophageal reflux (5/12 [42%]), developmental delay (3/12 [25%]) and recurrent lung infections (1/12 [8%]) were identified. ***Conclusions:*** Associated anatomic and genetic anomalies are common in omphalocele. Postnatal workup should include screening of these anomalies. Prognoses for isolated omphaloceles with no major postoperative complications are good in the long-term.

Key words Hong Kong; Omphaloceles; Outcome

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Introduction

Omphalocele, a rare congenital abdominal wall defect (Figure 1), has an estimated prevalence of 1 per 3000-4000 live births.[1] It has a high co-occurrence with other congenital anatomical or genetic anomalies. Survival rates of infants with omphalocele vary depending on the presence or absence of these associated anomalies. The purpose of our study is to review the perinatal characteristics, associated anomalies and clinical outcomes of omphalocele at a tertiary neonatal intensive care unit managing neonates with surgical problems in Hong Kong.

Methods

Study Cohort

All newborns diagnosed with omphalocele that were admitted to the neonatal intensive care unit at Queen Mary Hospital (QMH) were identified using the Clinical Data Analysis Reporting System (CDARS) from 1st January 2005 to 31st December 2014, using International Classification of Disease (ICD-9) coding number 756.7 for omphalocele. Cases included newborns that were born at our hospital or babies that were born at other hospitals and being transferred to us postnatally for further management. For cases with antenatal follow-up at our hospital, paediatric surgeon will be consulted and counselling will be given during antenatal follow-up. We collected both maternal and infant data including: gestational age (GA), birth weight, sex, race, mode of delivery, Apgar score (AS) at 5 minutes, concomitant diagnoses, age in days at first feeding, mechanical ventilation, any presence of pulmonary hypertension, total length of hospitalisation, and survival at discharge. Data were retrospectively collected and obtained from the medical records.

Definitions

Anomalies including omphalocele were diagnosed by the treating neonatologists. An omphalocele containing the liver is defined as a giant omphalocele. We classified anomalies as major congenital anomalies involving the cardiovascular, pulmonary, renal, gastrointestinal and genetic system. We defined first day of enteral feedings as the first day of life an infant received any type or amount of enteral nutrition. Duration of mechanical ventilation was defined as the total duration of mechanical ventilation (conventional or high-frequency) during the hospitalisation. Pulmonary hypertension was diagnosed by echocardiogram. Pulmonary hypoplasia was defined by the presence of narrow elongated chest and caudal declination of ribs on chest radiograph. Diaphragmatic dysfunction was documented using ultrasonography of diaphragmatic movement.

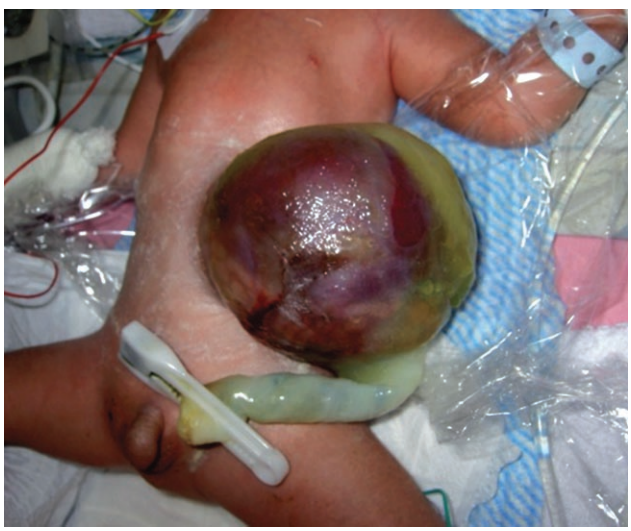

Figure 1 Omphalocele in newborn.

Statistical Analysis

The Fisher's exact test was used and a two-tailed p value of less than 0.05 was considered to be statistically significant.

Results

We identified 19 infants with omphalocele during the 10-year study period. Majority of cases (15/19 [80%]) were known antenatally. Median maternal age was 34 years. Infants were born with a median GA of 38 weeks with a median birth weight of 3140 g. There was a female predominance with male to female ratio of 1:1.7. Majority of cases (14/19 [74%]) were born via Caesarean section with a good AS at 5 min of life (all cases with AS ≥8). The decision for Caesarean section was dictated by obstetric indications only (independent on the presence of omphalocele). There was no rupture of omphalocele sac during spontaneous vaginal delivery. Twenty-one percent (4/19) required intubation at birth (Table 1).

Eleven out of 19 cases were giant omphaloceles (vs 8 non-liver containing omphaloceles). The mean diameter of the abdominal wall defect was 5 cm.

Associated anomalies were documented in 10 out of 19 infants (53%), among which congenital heart disease were identified most frequently (Table 2). Only one infant was diagnosed with >1 anomaly (gastrointestinal and renal anomalies).

Surgery was done in 17 cases (89%). Two patients died before surgery due to respiratory failure as a result of alveolar capillary dysplasia (ACD). Primary closure was achieved in 11 cases, while staged procedures (application of silo to the abdominal wall defect followed by delayed closure [Figure 2]) were carried out in 6 cases (Table 3).

Table 1 Demographics

	N = 19
Male	7 (37%)
Race	
Chinese	15 (79%)
White	1 (5%)
Other	3 (16%)
Inborn	16 (84%)
Caesarean section	14 (74%)
5-minute Apgar	
8	3 (16%)
9-10	16 (84%)
Intubation at birth	4 (21%)

Table 2 Associated anomalies

	N = 19
Cardiac	5 (26%)
Ventricular septal defect (VSD)	2
Atrial septal defect (ASD)	1
Patent Ductus Arteriosus (+ VSD)	1
Double-outlet right ventricle (DORV)	1
Pulmonary	2 (11%)
Alveolar capillary dysplasia (ACD)	2
Renal	2 (11%)
Duplex kidney	1
Vesicoureteric reflux	1
Gastrointestinal	1 (5%)
Imperforate anus, common cloaca	1
Genetic	1 (5%)
Beckwith-Wiedemann syndrome (BWS)	1

Table 3 Operation

Type of omphalocele	**N = 19**
Non-liver containing	8 (42%)
Primary closure	7
Died before operation	1
Liver-containing	11 (58%)
Primary closure	4
Silo followed by delayed closure	6
Died before operation	1

Median age at first operation (either primary closure or application of silo) was on day 1 of life, while delayed closure in staged operations was carried out at a median age of 8 days of life (range 7 to 9 days of life). Infants with non-liver containing omphaloceles were more likely to have primary repair compared to those with liver-containing ones (7/8 [88%] vs. 4/11 [36%], p=0.03). First feeds were started at a median postnatal age of 8 days.

A total of 4 infants were diagnosed with pulmonary hypertension, 2 cases received both inhaled nitric oxide and sildenafil as pulmonary vasodilating agents. These 2 cases were subsequently diagnosed to have ACD as post-mortem finding. Pulmonary hypoplasia and diaphragmatic dysfunction were diagnosed in 26% (5/19) and 11% (2/19) infants respectively. Giant omphaloceles were more likely to have pulmonary hypoplasia compared with non-giant types (p=0.04). Median duration of mechanical ventilation was 4 days (mean duration of mechanical ventilation for giant omphaloceles was 17.5 days vs 1 day for non-liver containing omphaloceles).

Overall survival rate at discharge was 84% (16/19). Survival rate at discharge was 100% for isolated omphalocele without other congenital anomalies. Mortality rate for giant omphalocele was 18% (2/11). Two cases died on day 1 and day 15 of life as a result of the lethal pulmonary condition of ACD. One case died on day 50 of life because of midgut volvulus due to intra-abdominal adhesion requiring massive small bowel resection. The median length of hospitalisation among survivors was 23 days (range 7 days to 1 year and 4 months). Majority of patients (12/16 [75%]) can be discharged within the first 60 days of life (range 7 days to 54 days).

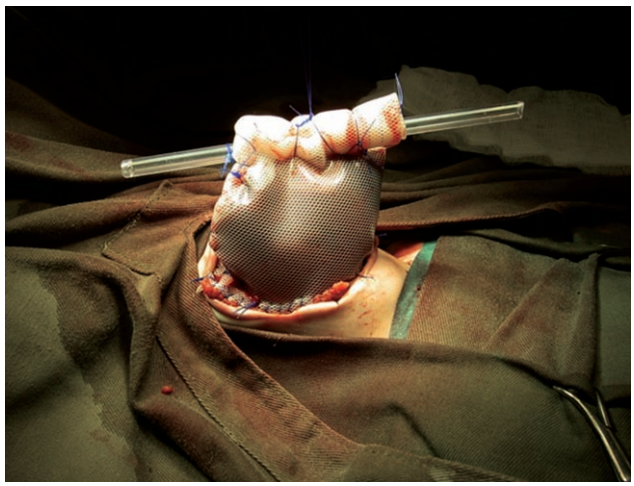

Figure 2 Staged repair of omphalocele.

Postoperative complications occurred in 6 patients (6/17 [35%]), with ventral hernia being the most common complication (total 3 cases with ventral hernia, 2 out of 3 cases underwent primary repair, 1 out of 3 cases with staged operation) (Table 4). Among the 16 survivors, 4 patients were lost to long-term follow up. Median duration of follow up was 5 years and 4 months (range 10 months to 10 years and 2 months). Long-term medical problems including failure to thrive (6/12 [50%]), gastroesophageal reflux (5/12 [42%]), developmental delay (3/12 [25%]) and recurrent lung infections (1/12 [8%]) were identified (Table 5). Clinical characteristics and outcomes of our case series were summarised in Table 6.

Table 4 Postoperative complications

	N = 17
Hernia	3 (18%)
Adhesion	2 (12%)
Wound infection	1 (6%)

Table 5 Long-term complications

	N = 12
Failure to thrive (FTT)	6 (50%)
Gastroesophageal reflux (GER)	5 (42%)
Developmental delay	3 (25%)
Recurrent lung infections	1 (8%)

Table 6 Summary of case series with omphalocele

Patient	**Sex**	**Type of omphalocele**	**Associated anomalies**	**Type of operation**	**Postoperative complications**	**Long-term complications**
1	F	Non-liver containing	No	Primary repair	Nil	No long-term follow-up
2	M	Non-liver containing	BWS	Primary repair	Nil	Nil
3	F	Giant	VSD	Staged	Hernia	GER, FTT, developmental delay
4	F	Giant	VSD, PDA	Staged	Nil	GER, FTT
5	F	Giant	VSD	Staged	Wound infection	GER, FTT
6	F	Non-liver containing	No	Primary repair	Hernia	Developmental delay
7	F	Non-liver containing	Imperforate anus, common cloaca, duplex kidney	Primary repair	Adhesion	No long-term follow-up
8	M	Giant	No	Staged	Nil	FTT
9	M	Non-liver containing	ACD	Passed away before operation	N/A	N/A (passed away)
10	M	Giant	ASD	Staged	Nil	GER, recurrent lung infections, FTT, developmental delay
11	F	Giant	No	Primary repair	Hernia	Nil
12	F	Giant	Vesicoureteric reflux	Primary repair	Nil	FTT
13	F	Giant	No	Primary repair	Nil	GER
14	M	Giant	No	Staged	Nil	Nil
15	F	Non-liver containing	No	Primary repair	Nil	No long-term follow-up
16	F	Giant	DORV	Primary repair	Adhesion, midgut volvulus	N/A (passed away)
17	F	Non-liver containing	No	Primary repair	Nil	No long-term follow-up
18	M	Non-liver containing	No	Primary repair	Nil	Nil
19	M	Giant	ACD	Passed away before operation	N/A	N/A (passed away)

BWS: Beckwith-Wiedemann syndrome; VSD: Ventricular septal defect; PDA: patent ductus arteriosus; ACD: alveolar capillary dysplasia; ASD: Atrial septal defect; GER: gastroesophageal reflux; FTT: failure to thrive; DORV: double-outlet right ventricle

Discussion

We described the perinatal characteristics, associated anomalies and outcomes of a cohort of infants with omphalocele from a 10-year period. In previously reported population-based studies, the occurrence of omphalocele appears to be more common in women at the extremes of reproductive age (<20 or >40 years of age).[2] Such observation was not detected in our study. The incidence of omphalocele in Hong Kong is not available as we are only one of the neonatal surgical units receiving cases in Hong Kong, and we do not have the number of prenatal cases and deliveries in other referring hospitals. There were a total of 15 cases of termination of pregnancies (TOP) and 3 spontaneous miscarriages related to omphalocele (with or without other congenital anomalies) during the study period (unpublished data obtained from personal communication with Dr. A Kan, Tsang Yuk Hospital Prenatal Diagnostic Clinic).

The proportion of neonates with co-occurring anomalies in our study (53%) was in range of other studies.[3,4] Our finding that ventricular septal defects, atrial septal defects and patent ductus arteriosus were the most common congenital heart defects associated with omphalocele is also consistent with others.[5] However, in this study we find a higher incidence of ACD, which is not reported in other studies. The need for use of both inhaled nitric oxide and sildenafil to manage refractory pulmonary hypertension should raise the clinical suspicion of ACD. Only one case (1/19 [5%]) was confirmed to have genetic disease (Beckwith-Wiedemann syndrome) in our series, this proportion is lower than other studies in the existing literature (15-17%).[5,6] A smaller proportion of cases with underlying genetic condition in our study population is probably related to the fact that more parents opted for TOP when genetic conditions were diagnosed antenatally. During the study period, 8 fetuses with omphalocele were diagnosed to have chromosomal abnormalities and subsequently underwent termination of pregnancy or spontaneous miscarriage (unpublished data obtained from personal communication with Dr. A Kan, Tsang Yuk Hospital Prenatal Diagnostic Clinic).

The overall survival rate at discharge in our study was 84%, with 100% survival rate among isolated cases, which is comparable to other international studies.[5,6] Mortality in our series were mainly contributed to the presence of lethal associated congenital anomalies i.e. ACD and major postoperative complication i.e. midgut volvulus resulting in massive gut resection with short gut syndrome not compatible with life. Majority of patients (12/16 [75%]) can be discharged within the first 60 days of life. Causes of prolonged length of hospitalisation included chronic lung disease secondary to lung hypoplasia, heart failure due to underlying congenital heart condition or social reason (await placement). With absence of associated anatomic/chromosomal anomalies and major surgical complications, the prognosis of infants with omphaloceles is favourable in the long term.

The data obtained from this study provide an important local source of information for the antenatal and postnatal counselling of parents whose infants were diagnosed to have omphaloceles. However, the data were collected from one single centre in Hong Kong with a small number of subjects only, rather than from population-based data, making establishment of statistically significant results difficult. We also did not have comprehensive data on the antenatal finding of omphalocele especially on the measurement of ratios e.g. fetal head circumference or abdominal circumference to size of omphalocele defect, to correlate with postnatal outcomes. We could overcome the fore-mentioned limitations by establishing a territory-based registry including all cases in Hong Kong and having more collaboration with our obstetricians on the antenatal evaluation of omphaloceles.

Declaration of Interest

None.

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