

# The Long-Term Outcome of Abdominal Wall Defects Exomphalos & Gastroschisis Retrospective Single Centre Study Children Hospital at Westmead, Sydney, Australia

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## Abstract

**Objective:** To examine the natural history and detailed outcome of diagnosed cases of abdominal wall defects that admitted to The Children`s Hospital at Westmead.

**Methods:** This was A retrospective single centre study of all cases of abdominal wall defects admitted between the 1<sup>st</sup> of October 1998 and the 1<sup>st</sup> of January 2007 to The Children`s Hospital at Westmead. The study had been conducted through a retrospective review of the neonatal, pediatric surgery records and subsequent follow-up information of all cases of Omphalocele and Gastroschisis diagnosed.

**Results:** Ninety cases with anterior abdominal wall defects were admitted. Majority of the cases were born at Westmead Hospital. There were 25 neonates with Gastroschisis (27 females, 28 males), and 35 neonates with Exomphalos (15 females, 20 males). Eighty-one neonates had prenatal ultrasound diagnosis. Some neonates had associated congenital anomalies which results in five deaths (4 Exomphalos, 1 Gastroschisis). Post operative complications were common and the postoperative hospital stay was often lengthy. Twenty-five patients out of the ninety (15 Gastroschisis, 10 exomphalos) required further operations which were related to their primary diagnosis of Gastroschisis or Exomphalos, mostly for abdominal wall hernias.

**Conclusions:** Long-term outcome was favourable in majority of cases with abdominal wall defects and mortality substantially happened in neonates with associated anomalies. Reoperations were necessary in those patients who had postoperative hernias.

**Keywords:** Abdominal wall defect, Gastroschisis, Exomphalos, Omphalocele.

## Introduction

Exomphalos is a prolapse of intraabdominal organs into the basis of the umbilical cord and is covered by a membrane consisting of peritoneum and amnion. The Exomphalos sac might contain the small bowel, colon, stomach and liver. The defect is thought to be caused by a rotational anomaly of the embryologic bowel between 6-12 weeks of gestation. Associated anomalies are seen in about 60% of fetuses with Exomphalos and those fetuses have an increased risk of chromosomal abnormalities (8), (16) and (11).

Gastroschisis is an abdominal wall defect occurring mostly to the right of the umbilicus which is characterized by the absence of a sac covering the prolapsed organs. Almost always one can see extracorporeal small bowel and colon floating in the amniotic fluid, a prolapse of liver and stomach is rarely seen.

Gastroschisis is supposed to be caused by vascular disruption eg disruption of the right omphalomesenteric artery (29). Others believe that Gastroschisis occurs after in utero rupture of a hernia of the umbilical cord (23) and (34).

Before 1970, Exomphalos was the commonest of the abdominal wall defects; it is now the second after Gastroschisis. The overall incidence is 1 to 2.5 per 5000 live births (46) with a male predominance. Gastroschisis has become the most common of the abdominal wall defects over the past 40 years (41, 47) this may be related to the increased incidence of prematurity and the increased survival of premature infants in general, or to the fact that it was not differentiated from the ruptured Exomphalos until the 1970s (15, 48). The incidence is about 4 to 5 per 10,000 live births (43) with a male predominance (49).

## Subjects and Methods

This was a retrospective single centre study of all cases of abdominal wall defects (Exomphalos and Gastroschisis, ICD-10 codes Q79.2 and Q79.3), which had been admitted between the 1<sup>st</sup> of October 1998 and the 1<sup>st</sup> of January 2007 to The Children's Hospital at Westmead. The study had been conducted through a retrospective review of the neonatal, pediatric surgery records and subsequent follow-up information of all cases of Omphalocele and Gastroschisis diagnosed in a 9-year period in The Children's Hospital at Westmead.

The data were retrospectively collected from the patient's hospital charts, medical records data and power chart had been used as references for the collection of data and follow-up information. The data which had been reviewed included: the antenatal sonographic diagnosis, gestational age which expressed in complete weeks and days, maternal details (such as mother age, smoking, drug abuser), sex, birth weight, delivery mode (caesarean section or vaginal delivery), any associated anomalies, the type of closure for the defect, any postoperative complications, the length of intubation (respiratory support), the total parenteral nutrition supplement (TPN), the time to resume enteral feeding and the length of hospital stay. Follow-up of affected infants was carried out by a review of pediatric surgery records and follow-up letters.

There was no confusion about the diagnosis because the patients were been already diagnosed, and the cases of ruptured exomphalos were diagnosed as ruptured exomphalos and had not been mistaken for a Gastroschisis. Majority of the cases were born at Westmead hospital (75% of Exomphalos cases and 84% of Gastroschisis cases) and transferred within first two hours to The Children's Hospital at Westmead for further surgical management. A variety of surgical techniques had been used in the treatment: painting with iodine, silo bag application, ward reduction, single stage closure and multiple staged closures. Postoperative complications had been encountered in 30 patients (21 Gastroschisis, 9 Exomphalos). The hospital's computer system had been accessed in order to get the available follow-up information for the patients. The duration of follow-up ranged from 6 months to 10 years. The follow-up information was available for seventy patients (40 Gastroschisis and 30 Exomphalos).

## Results

### Gastroschisis

During the study period 55 cases of diagnosed Gastroschisis had been managed in the Children's Hospital at Westmead. 25 males (51%) and 27 were females (49%), the prenatal diagnosis using ultrasound scan was made in forty nine cases (89%). The mean age for the mothers of babies with Gastroschisis was 25.5 years (range 15 to 38 years). Seven percent of the mothers (4/55) were heavy smokers during pregnancy and seven percent (4/55) were drug abusers (one mother was abuser for heroin, one for cannabis and the two others were addicted to methadone). Out of the 55 children 22 patients (40%) were born by spontaneous vaginal delivery, of which six cases had not been prenatally diagnosed. The other 33 cases (60%) were delivered by caesarean section for a number of obstetric reasons; emergency caesarean was performed in twelve cases because of fetal distress in seven cases, reduced fetal movements with onset of premature labour in four cases and because of dilated bowel in one case. While elective caesarean was done in the remaining 21 cases because of breech presentation, Gastroschisis. In all of those children who born by caesarean section the prenatal diagnosis had been made. The mean gestational age was 36 weeks plus three days (31 to 40 weeks range), forty five percent of cases were premature. The mean birth weight of babies was 2.5 kilograms (range from 1.2 to 3.8kg).

Associated anomalies had been diagnosed in thirteen patients; intestinal atresia was the commonest anomalies in six patients (11%), one of the children had colonic atresia and type 1 classical choledochal cyst (hepatomegaly, splenomegaly and jaundice), the second most common associated anomalies was genitourinary anomalies, which were seen in five patients (9%) including inguinal hernia in two cases, undescended testis in two cases and hypospadias in one case. Musculoskeletal anomalies were found in four cases (7%); two children had bilateral congenital acetabular dysplasia, one child had arthrogryposis (arthrogryposis multiplex congenita (AMC), is a rare congenital disorder which is characterized by multiple joint contractures and can include muscle weakness and fibrosis) and the last one had haemangioma to his right little toe.

The cardiovascular anomalies had been seen in three cases (6%). First child was preterm male who had PDA, ASD, hypoplastic right heart and pulmonary valve atresia. The second child was a full-term female who delivered by caesarean section; she had patent foramen ovale and pulmonary artery hypoplasia. The last child was a full-term male who delivered vaginally at Westmead hospital, and he had pulmonary hypertension.

In regard to the surgical management, the primary closure of the abdominal wall defect had been achieved in 27/55 (49%) of the cases, while staged closure after silo application was done in thirty three percent of the cases (18/55), and Gastroschisis reduction at ward was successful in the other ten cases (18%).

### **Exomphalos**

35 cases of diagnosed exomphalos had been admitted to the surgical unit at the Children's Hospital at Westmead during the study period. 20 cases (57%) were males, and fifteen cases (43%) were females. Prenatal diagnosis of exomphalos by ultrasound was done in 31 cases. The average maternal age at birth was 30 years (range from 17 to 42 years). Two of the mothers (6%) were heavy smokers during pregnancy. Two of the cases (6%) were induced pregnancies (in vitro fertilization IVF). Out of the 35 patients fifteen patients were born by spontaneous normal vaginal delivery (43%) and two of them were not prenatally diagnosed, the remaining children (57%) were born by caesarean sections for many reasons. Emergency caesarean section were done in 8/35 (23%) because of premature labour with reduced fetal movements, and in three patients (9%) because of fetal distress and in one case because of twin pregnancy with intra-uterine growth retardation (IUGR). Elective caesarean section was indicated for the remaining patients for breech presentation, prolonged rupture of membranes, induced pregnancy and because of exomphalos major. The prenatal ultrasound diagnosis was done in all of the patients who delivered by caesarean section. The mean birth weight was three kilograms (range from 940 grams to 4.4kg). The mean gestational age was 35 weeks (29 weeks to 40 weeks). Thirteen patients (37%) out of the 35 were premature.

35 of the 35 patients (71%) had associated congenital anomalies. The most common congenital anomalies were the congenital heart diseases which had been encountered in 16/35 (47%). The cardiac anomalies included patent ductus arteriosus (PDA) in 10/35 (29%) patients either isolated or associated with other cardiac defects, four patients had pulmonary hypertension, one patient had tetralogy of fallot (The combination of a VSD with Pulmonary Stenosis, with the Aorta "Overriding" the VSD and with RV Hypertrophy). One patient had pentalogy of Cantrell (Diaphragmatic and ventral hernias, hypoplastic lung, tetralogy of fallot, cleft palate, omphalocele). Large ASD (atrial septal defect) found in two cases. Ventricular septal defects were found in three cases. One patient had double outlet left ventricle with hypoplastic left heart; complete atrio-ventricular canal had been diagnosed in two patients, two of the patients had coarctation of aorta (COA). Patent foramen ovale was found in three patients and one patient had left ventricular dysfunction.

Genitourinary congenital anomalies were found in ten cases (29%). Inguinal hernias were diagnosed in five patients (11%), four of them had right inguinal hernias while the last one had bilateral inguinal hernia. Undescended testis was found in three children one of whom had bilateral undescended testis and the other two had left undescended testis. One patient had bilateral retractile testis and the last patient had an extrarenal pelvis.

Gastrointestinal anomalies had been encountered in six children (18%). Meckel's diverticulum was found in three of them (9%), two patients had intestinal atresia, one of those who with atresia had right abdominal cyst. The last patient had exomphalos minor with enteric fistula (patent vitellointestinal duct).

The musculoskeletal anomalies were seen in two cases (6%), one patient was preterm female and she an infant of diabetic mother with sacral agenesis, bilateral hip dislocation and spina bifida. The other patient was a premature male with scoliosis.

Ear deformities encountered in two patients (6%), one of whom was a preterm male with low set ear and he had associated laryngotracheal cleft. The second patient was a full term female and she had right ear deformity. Both of them had a negative hearing test.

Chromosomal abnormalities had been encountered in two patients (6%). One patient was diagnosed as trisomy 21 (Down syndrome) and the other child had trisomy 11 (Emanuel syndrome).

Beckweith-wiedmann was diagnosed in eight cases (23%), also known as Exomphalos-macroglossia-gigantism syndrome [EMG syndrome] and Wiedemann-Beckwith syndrome [WBS] Pediatric overgrowth disorder involving a predisposition to tumour development. The clinical presentation is highly variable; some cases lack the hallmark features of Exomphalos, macroglossia, and gigantism. Abdominal wall defects common, as well as visceromegaly including liver, spleen, pancreas, kidneys, and adrenals.

**Table1:** Management.

	Gastroschisis	Exomphalos
Average initial	34 days	21 days
Surgical treatment		
1ry closure of abdominal wall	27/55 (49%)	25/35 (71%)
Staged closure (after silo)	18/55 (33%) 10/55 (18%)	3/35 (9%) 2/35 (6%)
Ward reduction		1/35 (3%)
Painting No treatment		4/35 (11%)
TPN		
Administered to	52/55 (95%)	22/35 (67%)
Respiratory support duration	4 days	6 days
Time for oral feeding establishment	18 days	5 days

**Table2:** Postoperative complications.

	Gastroschisis	Exomphalos
Sepsis	10/55 (18%)	1/35 (3%)
Metabolic acidosis	8/55 (15%)	
Oral feeding intolerance	4/55 (7%)	1/35 (3%)
Postoperative jaundice	4/55 (7%)	
Respiratory complications	2/55 (4%)	4/35 (11%)
Wound infection	1/55 (2)	3/35 (9%)
Wound dehiscence	2/55 (4%)	
Opioid dependency	2/55 (4%)	1/35 (3%)
Hypoproteniemia	2/55 (4%)	
2ry hypothyroidism		1/35 (3%)
Rotaviral gastroenteritis	2/55 (4%)	
Enterocolitis	1/55 (2%)	
Venous thrombosis 2ry to cath-	1/55 (2%)	

**Table 3:** Follow-up information.

	Gastroschisis	Exomphalos
Available information	40/55	30/35
Average follow-up period	19 months	38 months
Medical problems		
Asthma	3/55	2/35
Neurological, ENT	1/55	4/35
Gastroenterology	1/55	
Cardiology	1/55	
Delayed mile stones	1/55	
Below 3 <sup>rd</sup> centile weight	1/55	1/35
mortality	1/55 (2%)	4/35 (11%)

## Discussion

In our study 80 patients out of 90 had ultrasound, exomphalos was detected prenatally in 31/35 (89%) and Gastroschisis in 49/55 (89%). The pathogenesis or the etiological causes of abdominal wall defects and the predisposing risk factors are still a controversial. In the literature Gastroschisis had been linked to young maternal age (36-38). In our cases it is shown that the mothers of Gastroschisis patients were significantly younger than the mothers of Exomphalos patients (mean age for Gastroschisis cases mothers 24.5 years versus 30 years for exomphalos cases mothers). In the literature other risk factors for abdominal wall defects had been reported such as high maternal nicotine and the drug abuse (38). In our study 7% of the mothers in Gastroschisis were heavy smokers and other 7% were drug abusers (cocaine, methadone and cannabis), while in Exomphalos patients 6% of the mothers were smokers. In regard to the familial tendency to abdominal wall defects we showed no evidence for a familial recurrence because the family history was negative in all patients. The associated congenital malformations are more seen in patients with exomphalos than patients with gastroschisis (52, 55). In our study 47% of exomphalos cases were had cardiovascular anomalies in comparison to 3% of Gastroschisis patients. Chromosomal abnormalities and congenital syndromes are found in 4% of exomphalos cases 2% had trisomy 21 (Down syndrome) and 2% had trisomy 11. While Beckwith-Wiedemann syndrome had been encountered in 23% of Exomphalos cases. Gastroschisis usually was an isolated defect when compared to exomphalos and has the best outcome.

The follow up information were available in 73% of Gastroschisis cases and in 86% of Exomphalos cases with an average follow up period of about one and half year for Gastroschisis and three years for Exomphalos. The data collected included the surgical follow up regarding the surgical scar and the umbilicus, gastrointestinal problems, physical and intellectual growth.

The mortality among the abdominal wall defects patients, as described in the previous literature, had been markedly decreased in recent years. This improvement had been seen after the improvement in prenatal diagnosis technique and the advanced neonatal care and surgical practices. The mortality in exomphalos group was mainly because of the associated congenital anomalies in our study, and this is agreed with what had been published in the previous literature.

## Conclusions

- The long-term outcome was favourable in majority of cases with abdominal wall defects.
- The direct closure is the treatment of choice, but when there is evidence of raised intraabdominal pressure the staged closure after synthetic materials application will be the best way of management.
- The advances in neonatal care and the administration of total parenteral nutrition had significantly improved the outcome among the abdominal wall defects patients.
- Post-operative wound and gastrointestinal problems usually improved with the time and no long term side effects in the majority of cases.

- The mortality to large extent happened in patients with other associated congenital anomalies.
- The physical and mental growth is expected to be normal, unless there are other associated chromosomal or congenital anomalies, which could be severe, but the abdominal wall defects themselves are not expected to impair the life quality.

## Ethics Consideration

Ethics approval had been obtained (LNR/12/SCHN/178).

## Data Analysis

The statistical analysis had been carried out by Microsoft Excel & SPSS version 20. The data had been summarised by using means, standard deviations and percentiles.

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