Pediatr Surg Int (2009) 25:413–416
DOI 10.1007/s00383-009-2357-3

ORIGINAL ARTICLE

Neonatal survival of prenatally diagnosed exomphalos

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Accepted: 11 March 2009 / Published online: 26 March 2009

Abstract
Purpose Exomphalos is a midline defect, with a viable sac composed of amnion and peritoneum containing herniated abdominal contents with an incidence of about 1 in 4,000 live births. Associated major abnormalities can be karyotypic, syndromic or structural in up to 70% of cases. The aim of this study is to determine the factors that influence survival of antenatally diagnosed exomphalos.
Methods All antenatally diagnosed and postnatally confirmed exomphalos registered with our fetal medicine unit, during 2002–2007, were reviewed. Both prenatal and postnatal outcomes were analysed.
Results Of 88 cases identified with exomphalos, 85 were prenatally diagnosed. Fifty-five of them died in utero (45 terminations, 5 spontaneous abortions and 5 still births). There were 33 live births (37.5%), 7 of which were premature (30–35/40 gestation). Five babies died before coming to surgery (all with major exomphalos as well as abnormal karyotype) while 28 were operated upon. Fourteen cases with minor exomphalos, all isolated, were primarily closed and all survived to discharge. Of 14 babies with major exomphalos, 4 were closed primarily. Nine required silo formation and six successfully underwent secondary closure (one of which had a prenatal diagnosis of giant ruptured exomphalos). Three died before closure, two from sepsis and multi-organ failure, and one from an undiagnosed tracheo-oesophalgeal cleft. All three deaths had antenatally diagnosed giant ruptured exomphalos and were less than 34/40 weeks gestation. One baby was managed conservatively with antiseptic solution applied to the sac and left to heal by secondary intention. There were 17 cases of isolated exomphalos (with no other structural abnormalities), all of which survived.
Conclusion Antenatal diagnosis of exomphalos is 96% sensitive. Severe karyotypic and structural abnormalities were present in all intra-uterine and early postnatal deaths. Overall survival to discharge was 28%. Both minor and isolated exomphalos carried a good prognosis. Isolated exomphalos was a better prognostic factor than severity of the exomphalos itself. Ruptured giant exomphalos were associated with a poorer outcome especially in premature babies.

Keywords Exomphalos · Omphalocele · Antenatal diagnosis

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Purpose

Exomphalos is a congenital defect of the fetal abdominal wall present in approximately 1 in 4,000 live births [1]. It is thought to arise when the midline layers of muscular and aponeurotic tissue fail to fuse leaving the fetus with a characteristic midline defect usually near the insertion point of the umbilical cord, with a viable sac composed of amnion and peritoneum containing herniated abdominal contents. It is differentiated from gastroschisis by the presence of the peritoneal sac, although this may not be intact. Unlike gastroschisis, it is associated with various major karyotypical and morphological abnormalities. These include trisomy 13, 18 and 21, Beckwith Wiedemann syndrome (macroglossia, gigantism, exomphalos) and Pentalogy of Cantrell (sternal, pericardial, cardiac, abdominal wall and diaphragmatic defect). Cardiac,

gastrointestinal or renal abnormalities such as septal defects, hydronephrosis and intestinal atresias are noted in up to 80% of cases [1].

Exomphalos can be classified into major and minor, depending on the size of the defect, the presence of herniated, solid abdominal organs and into intact or ruptured, depending on whether the covering membrane has broken [2, 3]. Most minor defects are closed primarily, whereas there is more controversy regarding closure of major and ruptured exomphalos [4, 5].

Antenatal diagnosis of exomphalos was first encountered in 1978 [6]. This resource, now widely used in the developed world, allows prenatal counselling of the parents, and planning for delivery in a tertiary centre where paediatric surgeons and neonatal intensive care support is available—all shown to increase survival [7]. Routine antenatal anomaly scans are performed at around 20 week gestation in order to detect a variety of malformations. Reported sensitivity of ultrasound scanning (USS), to diagnose exomphalos, is about 75% with significant regional variation.

Aim

The aim of our study was: (1) to determine the diagnostic accuracy of prenatal diagnosis and (2) to determine the factors that influence survival of antenatally diagnosed exomphalos.

Methods

There are 7,000 deliveries per year at the John Radcliffe Hospital in Oxford. The details of all regional neonatal procedures are entered into a Congenital Anomaly Register for Oxfordshire, Berkshire and Buckinghamshire (CAROBB) [8]. This was searched retrospectively for antenatally diagnosed and postnatally confirmed abdominal wall defects between 2002 and 2007 (inclusive) and cross checked with the Oxford Paediatric Surgical Database.

Exomphalos made up 88 cases when compared with 61 cases of gastroschisis over the same time period. However, the incidence of gastroschisis had increased twofold over the last 3 years of the study period. Pre- and postnatal outcomes of exomphalos were recorded.

We defined minor exomphalos as a defect <5 cm, and major exomphalos as a defect greater than 5 cm. Giant exomphalos was defined as the presence of the entire liver in the sac. Exomphalos was considered isolated if there were no morphological or karyotypical abnormalities identified.

The criteria for delivery was normal vaginal delivery at term and caesarean section recommended to patients in whom the risk of fetal liver damage was high during vaginal delivery. Primary closure was the preferred operative management for exomphalos and silo only used where primary closure was not feasible. Non-operative management was reserved for infants with giant exomphalos in whom even the use of a silo over a reasonable time would not allow for reduction of abdominal contents, and those babies with comorbidity.

Results

Eighty-eight cases of exomphalos were diagnosed over a 6-year period. Eighty-five of these were prenatally diagnosed using USS at nuchal scanning and at the 20 week anomaly scan. Three were postnatally diagnosed and all three were minor exomphalos similar to a cord hernia. There were 33 live births (37.5% of the total). Seven of these were premature (21% of live births) (Fig. 1). There were 55 prenatal deaths, 45 of these were elective terminations due to severe associated anomalies, 5 were spontaneous abortions and there were 5 still births. There were eight postnatal deaths all of which had major chromosomal anomalies. Five died prior to surgery, while three died after primary surgery (Fig. 1).

All 45 elective terminations had the prenatal ultrasound findings confirmed with fetal autopsies and where autopsies were declined, by fetal examination.

In this group of elective termination, the karyotype was abnormal in 27 cases (trisomy 18, 13, 16), normal in 14 cases, and 4 patients declined amniocentesis. The 14 patients with normal karyotype had multiple complex abnormalities, which included neurological abnormalities in six fetuses (spina bifida 2, enencephaly 3, Dandy Walker abnormality 1), pentalogy of Cantrell 3, multiple skeletal abnormalities with cleft lip and palate plus complex cardiac

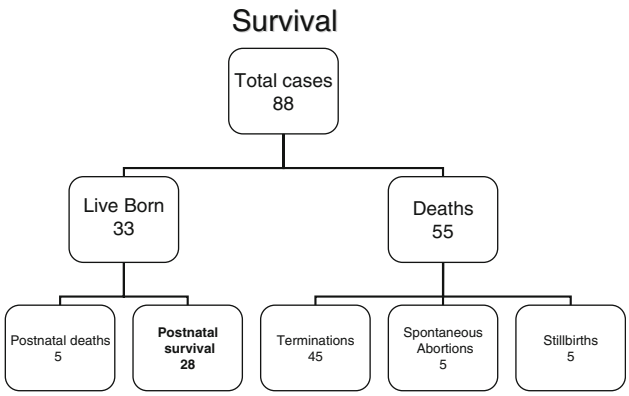

Fig. 1 Outcome of prenatally diagnosed exomphalos

abnormality in 2, body stalk abnormality in 2 and one isolated giant ruptured exomphalos had a social termination. The fetal anomalies confirmed on the four patients who declined amniocentesis include spina bifida in one, body stalk abnormality in two and enencephaly and in one fetus.

Of the 28 neonates who survived, 14 had minor exomphalos, all of which were isolated and all of them survived primary closure (Fig. 2).

Fourteen had major exomphalos, four of which were closed primarily. Nine had a silo bag placed and one required conservative treatment. Of the nine neonates with silos, six came to secondary closure, but three died before closure (Fig. 3).

These three postoperative deaths were in neonates with antenatally diagnosed giant ruptured exomphalos, born at less than 34/40 gestation.

One died from an undiagnosed tracheo-oesophageal cleft with major chromosomal anomalies, and the two others had sepsis and multi-organ failure.

Overall outcome of the 88 cases was 25 (28%) neonatal survival and 63 (72%) deaths. The deaths included 55 (87%) prenatal, 5(8%) postnatal but preoperative and 3 (5%) neonatal deaths postoperative (Fig. 4).

Discussion

Of the 88 cases of fetal exomphalos that presented to our department, all but 3 were diagnosed prenatally at nuchal scan or 20 week anomaly scan. This gives an in house sensitivity of 96.5%. There were no false-positives or false-negatives and there were no adverse fetal consequences from the diagnostic procedure for fetal karyotyping. The value of sonography to identify fetal abdominal wall defects has been recognised to enable forward planning to optimise management of the neonate and is now a well-established diagnostic tool [7, 9]. There are not much data

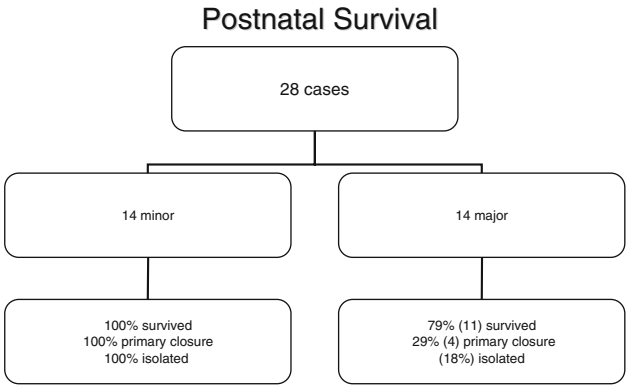

Fig. 2 Postnatal survival of exomphalos major and minor

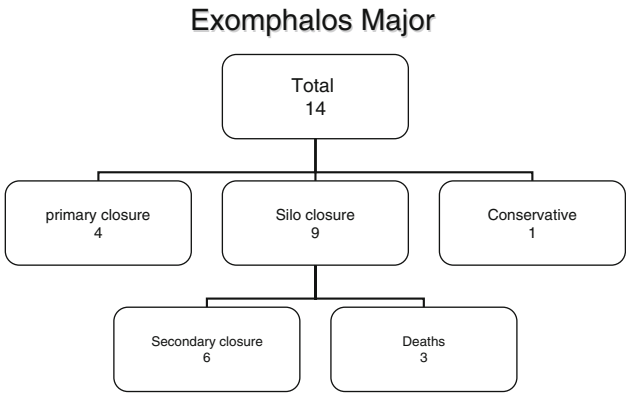

Fig. 3 Management of exomphalos major

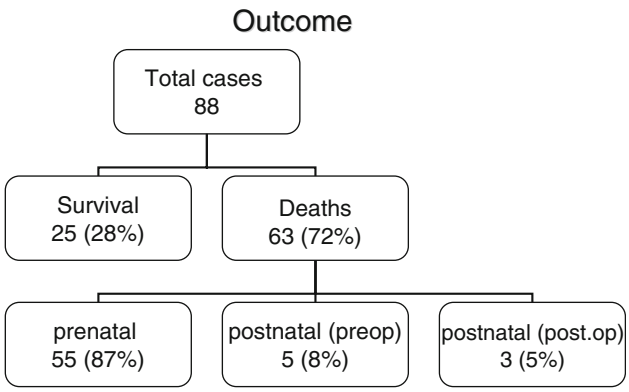

Fig. 4 Postnatal outcome of prenatally diagnosed exomphalos after surgical management

on the usefulness of non-invasive biochemical testing such as alfa fetoprotein as an adjunct to this.

Only 37.5% of the fetuses were live born. The majority (82%) were electively terminated and the rest were either spontaneous abortions or still births. There were also eight postnatal deaths of neonates with severe malformations. This is not an anomalous finding—other studies have reported similarly high in utero attrition rates [2, 10]. It highlights the importance of prenatal diagnosis when dealing with exomphalos. The spectrum of severity in terms of associated abnormalities is so wide that looking for seriously disabling karyotype and structural malformations is rigorous, since implications for both the parents and the fetus are significant and can cause much distress if not properly counselled.

Of the 28 neonates that came to surgery, half had primary closure of their defect without any postoperative sequelae. Eleven of these neonates had isolated exomphalos, i.e. no other defects which had a bearing on their relatively straightforward management and good outcome. Although the majority of our cases of minor exomphalos were isolated, the incidence of minor exomphalos with other karyotypical and structural malformations is not

negligible—reported to be 39% [1]. Nevertheless, our experience with major exomphalos showed that isolation is a good prognostic indicator as all our major isolated exomphalos cases had good outcomes.

Our poorest outcomes were in premature babies with ruptured giant exomphalos [2, 11, 12]. One had a trache-oesophageal cleft that was not compatible with survival so management of the exomphalos was secondary. The other two both had silos applied but were physiologically unstable and died of respiratory sepsis and multi-organ failure. This is probably more a reflection of the multiple co-morbidities associated with prematurity.

Conclusion

USS for the diagnosis of exomphalos yielded 96% sensitivity. Overall survival to discharge was 28%, with the highest mortality occurring prenatally. Neonates with minor and isolated exomphalos carried a good prognosis (100% survival). Isolated exomphalos is a better prognostic factor than severity of the exomphalos and finally giant ruptured exomphalos in premature babies carried the worst outcome.

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