OUTCOME OF ANTENATALLY REFERRED CONGENITAL SURGICAL ANOMALIES - A PEDIATRIC SURGEON’S PERSPECTIVE -

DK Gupta, Shilpa Sharma, Malvika Gupta
New Delhi, India

ABSTRACT

Background: With improvements in investigative modalities and increasing incidence of antenatal diagnosis of surgical anomalies, the pediatric surgeon has the added responsibility of deciding the fate of the unborn life and counselling about the ultimate outcome of the surgical anomaly diagnosed relying on his experience.

Material and methods: The clinical antenatal record files of 91 cases of patients referred to the department of pediatric surgery over a period of 4 years were thus studied in detail. The data was analysed in terms of the gestational age at presentation, anomaly detected, antenatal advice given and their postnatal outcome. All the cases were evaluated on the basis of antenatal ultrasonography. The gestational age at presentation was 12-20 wks; 21-28 wks; 29-34 wks and 34 wks – term in 13(14%); 22(24%); 35(38%) and 21(23%) cases respectively. 33 (36%) were primigravida. 16 had a bad obstetric history. The distribution of cases according to diagnosis was Urological – 43(47%), GIT – 12(13%), Cranial –17(19%), Spinal – 10(11%), Thoracic- 7(8%), Cleft Lip-1 (1%) and Tumor –3 (3%). Two fetuses had more than 2 anomalies –2(2%). Medical termination of pregnancy was advised for 7(7.7%) patients. There was a male predominance with a male: female ratio of 2:1.

Results: Discrepancies between the antenatal and postnatal diagnosis were encountered in 8 (8.8%) cases. Postnatal surgery was done in 21(23%) cases. 40(43.9%) are in regular follow up and close observation. 7(7.7%) cases expired with and without surgery. Surgery is planned in future for 5 cases and delivery of 4 cases is awaited. 7(7.7%) cases have been lost to follow up.

Conclusions: Delivery of high-risk infants in regional centres improves neonatal surgical care. An improvement in antenatal care especially in high-risk pregnancies, prenatal diagnosis and management at tertiary care level centre is still required in developing countries.

Key words: antenatal diagnosis, congenital surgical anomalies, neonatal surgery

Antenatally diagnosed congenital surgical anomaly are usually first brought to the notice of the obstetrician who decides the fate of the foetus. Out of the 8,000 antenatal cases seen each year at the authors’ tertiary level care Hospital, about 5-6 % are found to have congenital anomalies. Of these 80 % are terminated at the level of the obstetrician. For surgically correctable congenital anomalies, the obstetrician may seek prenatal pediatric surgical consultation that may have a significant impact on the perinatal management of the fetus. Providing a valuable insight into the surgical management of anomalies allows fetal intervention when appropriate, delivery in an appropriate setting, by the safest mode of delivery, and at the gestational age appropriate to minimize effects of the anomaly [1]. The pediatric surgeon has to counsel the parents on his ability to successfully treat the identified anomaly [2].

Maternal Serum Alpha-fetoprotein (MSAFP) is now being carried out for all antenatal cases usually between 15-17 weeks of gestation in most tertiary centres. A high MSAFP has an accuracy of diagnosing 85% of the open neural tube defects and 75% of ventral wall defects. A low MSAFP is found in cases of Down syndrome and Trisomy 18. The accuracy in Down syndrome is 80-90% in women over 36 years and 60-70 % in women under 36 years.

The understanding of the natural history of many prenatally diagnosed surgical conditions has grown significantly in recent years. The purpose of this study was thus to evaluate the impact of pediatric surgical consultation on antenatally diagnosed congenital surgical anomalies and form guidelines on the antenatal advise to be given for the variety of anomalies diagnosed depending on the gestational age at presentation.

Aim of the study

To study the outcome of antenatally diagnosed congenital surgical anomalies referred to pediatric surgeons.

Correspondence:
DK Gupta, Professor and Head,
Department of Pediatric Surgery,
All India Institute of Medical Sciences,
Ansari Nagar, New Delhi-110029, INDIA.
Email: profdkgupta@gmail.com, devendra6@hotmail.com
Patients and method

The clinical antenatal files of 91 cases referred to the department of Pediatric Surgery between 2002-2005 were studied in detail. The data was analysed in terms of the gestational age at presentation, diagnosis, antenatal advice given and their outcome. All the cases were evaluated on the basis of antenatal ultrasonography.

Results

All patients had undergone a routine MSAFP test at 15-17 weeks of gestation. If the MSAFP values were found abnormal, the Triple test comprising of Maternal Serum Alpha-fetoprotein, HCG and Estriol was advised. These tests were found abnormal in 1-2 % cases that were then subjected to further tests like amniocentesis and chorionic villous biopsy. A routine ultrasonography was done at 12-14 weeks and then at 18-20 weeks for screening for congenital anomalies. The presence of a congenital correctable congenital surgical anomaly was an indication for soughing pediatric surgical opinion.

The gestational age at presentation was 12-20 wks; 21-28 wks; 29-34 wks and 34 wks – term in 13(14%); 22(24%); 35(38%) and 21(23%) cases respectively. 33 (36%)were primigravida. 16 had a bad obstetric history. There was a positive history of drug Intake in 7 patients. The maternal and paternal age at presentation is depicted in Table 1.

95 anomalies were identified in 91 cases, Urological – 43(47%), GIT – 12(13%), Cranial –17(19%), Spinal – 10(11%), Thoracic- 7(8%), Cleft Lip-1 (1%), Tumor - 3 and more than 2 anomalies –2 (Table 2). Four patients had both hydrocephalus and meningomyelocele. Medical termination of pregnancy was advised for 7 patients comprising 3 patients with posterior urethral valve and oligohydramnios and one each with hydrocephalus, diastematomyelia, bilateral polycystic kidney disease and neural tube defect. There was a male predominance with a male: female ratio of 2:1

Discrepancies between the antenatal and postnatal diagnosis were encountered in 8 (8.8%)cases. Four cases with a diagnosis of a surgical anomaly were found normal postnatally. These included one each of CDH, omphalocele, hydrocephalus and abdominal cyst. One case with an antenatal diagnosis of Mega Cisterna Magna turned out to be one of Turners Syndrome. A case of antenatal sacrococcygeal teratoma was a LSMMC. A case of CDH turned out to be CCAM and a case of CCAM was actually a bronchogenic cyst.

Seven cases were medically terminated. Surgery was done in 21(23%) cases. These included 6 thoracic, 2 MMC, 5 abdominal, 7 urological and 1 RMS cases

Surgery is planned in future for 5 cases; 3 urological, 1 omphalocele and 1 choledochal cyst. Thus 26 (28.9%) cases would ultimately be requiring surgery.

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40 are in regular follow up and close observation. One baby with antenataly detected meconium peritonitis delivered preterm with gross abdominal distension at birth due to formation of giant meconium cyst (Fig 1 A & B). The ribs were flared due to the gross abdominal distension causing respiratory compromise. Double barrel ileostomy was done which was followed by adhesinolysis and ileoileal anastomosis after 10 days. The baby required postoperative ventilation for 6 days to stabilize. An antenatally detected Neuroblastoma showed a persistent adrenal solid mass in the postnatal USG and CT Scan that disappeared after 1 month of life (Fig 2 A, B & C). The postnatal catecholamines were normal. Seven cases expired. Delivery of 4 cases is awaited. Seven cases have been lost to follow up.

<table>
<thead>
<tr>
<th>Mother (n= 91)</th>
<th>Father (n = 91)</th>
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<tbody>
<tr>
<td>18-20</td>
<td>10</td>
</tr>
<tr>
<td>21-24</td>
<td>27</td>
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<tr>
<td>25-28</td>
<td>42</td>
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<td>29-32</td>
<td>10</td>
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<td>33-38</td>
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Table 1: Parenteral age at time of conception

Table 2: Antenatally diagnosed congenital surgical anomalies

<table>
<thead>
<tr>
<th>Antenatal Diagnosis</th>
<th>N=95</th>
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<tbody>
<tr>
<td>Urological diagnoses 43(47%)</td>
<td></td>
</tr>
<tr>
<td>Pyelocaliectasia (PUJO)</td>
<td>27</td>
</tr>
<tr>
<td>Posterior urethral valve</td>
<td>9</td>
</tr>
<tr>
<td>Multi Cystic Kidney Disease</td>
<td>4</td>
</tr>
<tr>
<td>Poly Cystic Kidney Disease</td>
<td>2</td>
</tr>
<tr>
<td>Renal Agenesis</td>
<td>1</td>
</tr>
<tr>
<td>GIT – 12 (13%)</td>
<td></td>
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<tr>
<td>Esophageal Atresia</td>
<td>1</td>
</tr>
<tr>
<td>Meconium peritonitis</td>
<td>2</td>
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<tr>
<td>Duplication Cyst</td>
<td>1</td>
</tr>
<tr>
<td>Omphalocele</td>
<td>4</td>
</tr>
<tr>
<td>Gastrochisis</td>
<td>2</td>
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<tr>
<td>Choledochal cyst</td>
<td>2</td>
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<tr>
<td>Cranial – 17 (19%)</td>
<td></td>
</tr>
<tr>
<td>Hydrocephalus</td>
<td>12</td>
</tr>
<tr>
<td>Dandy Walker</td>
<td>2</td>
</tr>
<tr>
<td>Porencephalic Cyst</td>
<td>2</td>
</tr>
<tr>
<td>Mega cisterna magna</td>
<td>1</td>
</tr>
<tr>
<td>Spinal – 10 (11%)</td>
<td></td>
</tr>
<tr>
<td>Meningomyelocele</td>
<td>8</td>
</tr>
<tr>
<td>Diastematomyelia</td>
<td>2</td>
</tr>
<tr>
<td>Thoracic – 7 (8%)</td>
<td></td>
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<tr>
<td>CCAM</td>
<td>3</td>
</tr>
<tr>
<td>CDH</td>
<td>3</td>
</tr>
<tr>
<td>Lung Cyst</td>
<td>1</td>
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<tr>
<td>Tumor – 3 (3%)</td>
<td></td>
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<tr>
<td>Neuroblatoma</td>
<td>1</td>
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<tr>
<td>Sacrococcygeal teratoma</td>
<td>1</td>
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<tr>
<td>Rhabdomyosarcoma</td>
<td>1</td>
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<tr>
<td>Cleft Lip</td>
<td>1</td>
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<tr>
<td>Multiple (&gt;2) anomalies</td>
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Discussion

Improvements in screening and diagnostic techniques now allow many congenital anomalies to be diagnosed...
It is, however, not wise to submit all pregnant women to a barrage of investigations. Screening is necessary before specific invasive investigations are initiated. These include history, physical examination, MS-AFP screening, estriol and hCG screening, and a Level I ultrasonography scan. Once at-risk pregnancies have been identified, a multidisciplinary team approach is commenced and further studies including Level II ultrasonography, amniocentesis, chorionic villus sampling, or cordocentesis can be performed so that an accurate diagnosis is available [3]. Advances in maternal serum screening and second-trimester ultrasonography have resulted in more judicious use of amniocentesis and chorionic villus sampling [4]. Fetal MRI may provide a more detailed description and insight into fetal anatomy, pathology and etiology in cerebrospinal, retroperitoneal and in neck/thoracic which improves prenatal parental counseling and postnatal therapeutic planning [5].

The diagnosis of an antenatal surgical malformation allows (1) fetal intervention if facilities exist, (2) in utero transfer and planned delivery in a surgical center, and (3) antenatal counseling of likely prognosis and outcome. Delivery in a neonatal center facilitates provision of neonatal care from the time of birth, thus avoiding the risks of transport over long distances [2]. Antenatal counseling for fetal surgical malformations by specialist staff reduces the levels of parental anxiety [3,6]. Health care professionals must elicit each parent’s particular perspective, be cognizant of their professional influence, and actively support parents from the time of the antenatal diagnosis [7]. This influences them positively in coping with the anomaly [8].

This study being conducted at a referral centre, the percentage of congenital anomalies was high. Most of the cases that were referred before 20 weeks of gestation were terminated medically by the gynaecologist without referring the cases to the pediatric surgeon. These included anencephaly, neural tube defects, polycystic kidneys, gastrochisis and omphalocele where the decision is unequivocal. On the contrary these also included few cases of minor anomalies like unilateral multicystic kidney, unilateral hydronephrosis and cleft lip where the decision of a pediatric surgeon would be to continue the pregnancy. These were terminated on parenteral wishes, as they preferred to have a normal baby. When a severe abnormality is found in one twin, selective termination of pregnancy may be considered. This technique is safe in dichorionic twins, but hazardous in monochorionic pregnancies. Selective termination in dichorionic twins is safer in the first trimester, underscoring the need for early prenatal diagnosis in twins [9].

Discrepancies between the antenatal and postnatal diagnosis were encountered in 7 (7.7%) cases in this study. However the rate of precision of antenatal diagnosis has been reported as low as 23.8% in other series [10]. The highest rate of positive prenatal diagnosis has been reported for hydrocephalus (50%) and urinary tract anomalies (42.8%) [10]. On the vice-versa, many cases of congenital surgical anomalies are missed inspite of antenatal ultrasonography especially gastrointestinal atresia, congenital diaphragmatic hernia and abdominal wall defects [10].

The cases referred to the pediatric surgeon in this study included cases that had crossed the gestational age of medical termination of pregnancy implying that routine ultrasonographic screening for congenital anomalies is far from sufficient in the peripheral areas from where cases are being referred to higher centres at an advanced gestation age. However, inspite of advanced gestational age, early diagnosis facilitates assessment of the development of the affected organ, possible timely antenatal correction, team consultation regarding the time and mode of delivery and preparations for postnatal correction [11]. Such an approach significantly influences the perinatal outcome. In case of urinary tract abnormalities, antenatal diagnosis by ultrasonography even after 28 weeks’ gestation may permit early treatment of the asymptomatic newborn and reduce later renal damage [12].

Urological anomalies formed the most common group in this series (47%) followed by cranial and gastrointestinal defects. In a large series of 234 congenital anomalies the anomalies comprised of genitourinary (36%), thoracic (16%), intraabdominal...
(14.5%), abdominal wall (10.6%), neurological (9%), skeletal (6%), and head and neck (2.5%) defects; tumors 2.5% and twins 2.5% [1].

However in another series of 218 anomalies detected antenatally by conventional ultrasound, Neural tube defects and abdominal wall defects were the most common [10]. This discrepancy may be due to the difference in terminations advised by the obstetrician without seeking surgical consultation and the gestational age at presentation.

The necessity for an interdisciplinary team approach - obstetricians, pediatric surgeons, neonatologists, geneticists, neurologists, has been emphasized time and again [13].

A multidisciplinary antenatal diagnosis and management (MADAM) model has been shown to lead to alterations in fetal or perinatal management; 42% cases had an alteration in prenatal management, 18% had a co-ordination of postnatal management and 16% had the establishment of a new treatment guideline, or the modification of an existing one [14]. In all, perinatal management was altered in 75% of cases. A multidisciplinary approach model thus functions as a forum for exchange of up-to-date scientific information, development of evidence-based treatment protocols and continuity of care through the pre-, peri- and postnatal periods [14].

There are few surgical conditions for which operative intervention is mandatory in the immediate postnatal period with fair to good prognosis. These include esophageo-gastrointestinal conditions like esophageal atresia, duodenal atresia, jejunoileal atresias, colonic atresia, anorectal malformations, meconium peritonitis and gastrochisis. These should be delivered at the centre with neonatal surgical facilities. Some semi-emergency conditions like cystic diseases may be evaluated after some time postnatally. These include duplication cysts, mesenteric cyst and ovarian cyst.

Some conditions like neural tube defects if diagnosed after the gestational age of medical termination of pregnancy have to be evaluated for the neurological status soon after birth in the Indian scenario and surgical intervention is justified if a socially productive life can be insured to the patient. Antenatally diagnosed omphalocele has to be evaluated postnatally for associated congenital anomalies and intestinal obstruction. A conservative management may be offered if the baby passes meconium normally with necessary aseptic precautions.

It is estimated that genitourinary anomalies comprise 20% of all antenatally detected fetal anomalies, and pyelocaliectasia is the most common one. The most common uropathies are ureteropelvic junction obstruction, vesicoureteral reflux, posterior urethral valves, and primary obstructive megaureter. Antenatal diagnosis of urinary tract anomalies allowed immediate prophylactic treatment of urinary infections, and decrease the risk of severe complications previously observed [15]. Detection of antenatal dilatation of the urinary tract does not always indicate postnatal urinary tract obstruction or even a significant genitourinary anomaly.

Most cases improve spontaneously, representing a temporary physiologic impedance and do not require surgery. In a series of 197 newborns, with prenatal pyelocaliectasia, ultrasonic follow-up showed that pyelocaliectasia resolved in 97%, while 3% still presented it in the postnatal ultrasound. Surgery was performed in 2% children [16].

Fig 2- A: Antenatal ultrasonography at 37 weeks gestation delineating an echogenic lesion in left adrenal region suggestive of neuroblastoma,
B: C T scan on Day 2 showing a hypodense lesion,
C: Ultrasonography at 1 month of life showing disappearance of the mass.
pyelocaliectasia may thus be safely observed and surgical correction is indicated if infection sets in, a lump is palpable or renal compromise is documented on serial renograms and ultrasonography. Voiding cystourethrogram may be performed soon after birth if posterior urethral valves are suspected, later in other cases. However, in cases of intermittent renal pelvic dilatation, often an indirect sign of reflux, the need for a voiding cystogram is controversial [15]. Diuretic renogram with mercaptoacetyl triglycine (MAG 3) has been used to access renal function and efficiency of kidney drainage [15].

Prenatal diagnosis has been found to be correct in about 60% of the urinary tract anomalies [17]. In the remainder the diagnosis was either incomplete or incorrect. Errors in the diagnosis resulted from difficulties in the differentiation of dilated ureter, intestinal dilatation, or intraperitoneal cystic masses [17]. Congenital cystic adenomatoid malformation and congenital lobar emphysema are the commonest antenatally diagnosed lung malformations followed by pulmonary sequestrations and bronchogenic cysts. The natural history of parenchymal lung lesions such as congenital cystic adenomatoid malformation and pulmonary sequestration has been altered by the advent of antenatal ultrasonography. Initial reports were characterized by a high (about 30%) incidence of adverse features like hydrops and a poor outcome but reports today show that antenatally diagnosed “cystic lung disease” have an excellent prognosis in the absence of signs of severe fetal distress.

The need for surgery should be based on appropriate postnatal investigations (like CT scan), rather than on antenatal behavior [18]. All infants with a prenatal diagnosis require postnatal evaluation. Patients should be evaluated for associated disorders. The natural history is variable. Regression of the sonographic appearance with a prenatal diagnosis of congenital lung malformations has been observed [19]. Amniotic puncture may be required for cases with hydramnios. Emergency surgical resection may be required in the presence of mass effects. The risk of pulmonary compression, infection and malignant degeneration makes resection imperative, even in asymptomatic patients. Lobectomy has excellent outcome. Antenatal ultrasonography allows diagnosis and treatment before any complication and this is the main advantage [20]. Some authors however may support the conservative management of selected neonates with CCAM [21]. Prenatal ultrasound appears reliable in the detection of pulmonary abnormalities but the variety of conditions identified postnatally suggests that specific prenatal diagnoses and prognoses should be avoided; prenatal counselling and perinatal management should be adapted accordingly [22]. Upto 31% of CCAM have been found to have other types of pulmonary abnormality postnatally and in 15% the lungs remained apparently normal [22]. Antenatally diagnosed CDH turning out to be a CCAM as seen in the authors series has also been reported previously [2].

The mortality rate for congenital diaphragmatic hernia when diagnosed antenatally, varies with fetal age and with the presence or absence of hydramnios and degree of pulmonary hypoplasia. Mortality among prenatally detected cases of CDH has been reported to be paradoxically high (83%) [23]. Many cases that would never have been treated in the past because of death before referral and treatment for severe pulmonary hypoplasia not compatible with life are thus observed and sometimes treated. Nevertheless, lung development continues to be a determining factor for survival even when intensive treatment at birth is available. Response to therapy is unpredictable before birth and proposed antenatal treatment is still far from being a realistic option. The acceleration time/ejection time ratio by Doppler velocimetry obtained at the main branches of fetal pulmonary artery which is consistent throughout gestational age from 20 to 39 weeks has been shown to be an accurate parameter to predict the subsequent development of pulmonary hypoplasia and clinical outcomes with high positive and negative predictive values [24]. The Doppler waveform of normal fetal pulmonary artery showed a “spike-and-dome” Pattern and normal values of acceleration time/ejection time ratio from the right and left pulmonary arteries were 0.17 +/- 0.04 and 0.15 +/- 0.04 respectively. The prognosis has improved dramatically in recent years, primarily due to advances in neonatal and surgical interventions. Neonatal survival rates with an antenatal diagnosis now exceed 80% in some centers [25].

2.6% women have been found to have abnormal fetal central nervous system findings. Small cerebellar size was the most frequent anomaly, followed by isolated mild ventriculomegaly and isolated choroid plexus pathology [26]. Suboptimal neurodevelopmental outcome was found in 24% of children with isolated ventriculomegaly, 19% with small cerebellum and in 9% with choroid plexus pathology [26]. Transcerebellar diameter may have a role to play in respect to its developmental implication [26].

Gastrointestinal and abdominal wall anomalies always need operative intervention to ensure complete cure if found to be persistent in the postnatal ultrasound examination. Intact omphalocele can accurately be distinguished from gastrochisis by detection of a membranous sac covering the herniated viscera, by liver protruding from the abdomen, and by the frequent association of major structural anomalies. Gastrochisis is characterized by the absence of these findings and the presence of bowel loops floating freely in the amniotic fluid. Prenatal ultrasonography allows rational decisions about perinatal management by distinguishing between omphalocele and gastrochisis and by screening for associated anatomic defects. Karyotype analysis should be a part of the prenatal workup [27]. Infants with other associated defects or with birth weight less than 1500 gr. have poor outcomes and cesarean section may not be justifiable [28]. However, following prenatal diagnosis in larger infants with isolated defects, some advocate cesarean section while others advocate normal delivery near term [27,28]. Few authors advocate that scheduled cesarean delivery at 36 weeks for gastrochisis, after confirmation of fetal lung maturity, presents the infant to the pediatric surgeon under controlled conditions and shortens neonatal hospital stay [29].

Accurate prenatal diagnosis allows maternal transport and in a few cases may alter the timing or mode of
delivery [27]. Congenital duodenal obstruction is usually diagnosed by antenatal ultrasound between the fifth month of gestation and term [8]. Although surgical intervention occurred earlier in the neonatal period, the outcome of infants with duodenal obstruction was not changed by providing an antenatal diagnosis [8]. An antenatally diagnosed choledochal cyst has to be confirmed in the postnatal period with an ultrasonography and HIDA, Few cases with biliary obstruction may necessitate urgent cyst excision and hepaticojejunostomy [30]. Non-obstructed patients are best explored after the age of three months. Teratomas may be diagnosed prenatally as a mass by antenatal sonography or presence of polyhydramnios. Some germ cell tumors of the fetus and neonate have a better prognosis than others. Neoplasms with gastric teratomas have the best survival rates, and those with intraperitoneal, germ-cell tumors the worst. Fetuses with teratomas detected antenatally have 3 times the mortality rate compared with postnatally diagnosed neonates [31]. Antenatal neuroblastoma needs to be evaluated in the postnatal period for urinary catecholamines. Aggressive therapy may be indicated if high. Few cases resolve spontaneously but require regular follow up [32].

Antenatal pediatric surgical consultation may alter the obstetricians view point to a large extent. In one series, the decision to terminate was changed in 3.6%, the site of delivery was changed in 37% to facilitate postnatal evaluation and initiate immediate treatment and the mode of delivery was changed in 6.8% to prevent dystocia, hemorrhage into a tumor, as in sacrococcygeal teratoma, or to provide an emergency airway, as in cervical teratoma [1]. The timing of delivery may also be changed in up to 4.5% cases to avoid further damage to fetal organs in cases of obstructive uropathy, gastro-schisis, sacrococcygeal teratoma with high-output failure, and hydrocephalus [1].

To summarize, the pediatric surgeon has a very important role to play in the team for antenatal counseling of pediatric surgical anomalies. A blend of fetal and maternal interest should be kept in mind and the aim remains to provide the mother with a healthy newborn and counsel the parents about the best possible outcome given in a setting and helping them to decide the fate of the unborn child. The decision should not be made on a single ultrasonographic scan and repeated evaluation is helpful not only to buy time for the parents to adapt to a particular situation but also to take into consideration the high incidence of discrepancy reported. Interdisciplinary approach is essential and the obstetrician should not take the sole responsibility of deciding the fate of the fetus with antenatally diagnosed congenital surgical anomaly.

REFERENCES