Prenatally diagnosed surgical malformations - who should decide next?

WHO SHOULD DEAL WITH ANTENATAL DIAGNOSIS?

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 author’s tertiary level care Hospital, about 5-6% are found to have congenital anomalies. Of these 80% are terminated at the level of the obstetrician. These include anencephaly, neural tube defects, poly cystic kidneys, gastrochisis and omphalocele where the decision is unequivocal. On the contrary these also include few cases of minor anomalies like unilateral multicystic kidney, unilateral hydronephrosis and cleft lip where the decision of a paediatric surgeon would be to continue the pregnancy. In most of these cases, the termination is done on strong parental wishes, as they prefer to have only a normal and perfect baby. This means that only one-fifth (20%) are sent to seek the opinion of the paediatric surgeon.

The radiologist does the ultrasonography and makes the diagnosis. The geneticist is consulted for the outcome of medical diseases, chromosomal anomalies, in cases with bad obstetrical history and also for congenital surgical anomalies.

Out of the 91 cases seen at the Antenatal counselling clinic at AIIMS over the past 4 years, 85% patients were referred after 20 weeks of gestations, thus leaving only 15% at the stage of a possibility for medical termination of pregnancy. This is striking and depicts the need to improve the antenatal diagnosis and the role of the paediatric surgeon in the initial stages.

AN INTERDISCIPLINARY APPROACH

The necessity for an interdisciplinary team approach - obstetricians, radiologists, pediatric surgeons, neonatologists, geneticists, neurologists, has been emphasized time and again.[1] A multidisciplinary antenatal diagnosis and management (MADAM) model has been shown to alter the perinatal management in 75% cases,[2] thus functioning 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.[2]

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 or to provide an emergency airway as in a case with cervical teratoma.[3] The timing of delivery may also be changed in upto 4.5% cases to avoid further damage to fetal organs in cases of obstructive uropathy, gastrochisis, sacrococcygeal teratoma with high-output failure, and in fetuses with hydrocephalus.[3]

INVESTIGATIVE MODALITIES

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

If the MSAFP values are found abnormal, the Triple test comprising of Maternal Serum Alpha-fetoprotein, HCG and Estriol is advisable. Though these tests are found abnormal in only 1-2% cases yet suggest undertaking further tests like amniocenteses and chorionic villous biopsy. A routine ultrasonography at 12-14 weeks and then at 18-20 weeks is recommended for screening for congenital anomalies in the foetus.

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 aetiology in cerebrospinal, retroperitoneal, neck and thorax that improves prenatal parental counselling and postnatal therapeutic planning.[5]
IMPLICATIONS OF ANTENATAL DIAGNOSIS

The diagnosis of an antenatal surgical malformation allows (1) fetal intervention if indicated and for which if the facilities exist, (2) in utero transfer and planned delivery in a better equipped 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, hypothermia, infection and aspiration. Antenatal counseling for fetal surgical malformations by specialist staff reduces the levels of parental anxiety. Health care professionals can elicit each parent’s particular perspective, becoming cognizant of their professional influence and actively supporting parents from the time of the antenatal diagnosis. This helps them positively in coping up with the anomaly.

DISCREPANCIES IN DIAGNOSIS

Discrepancies between the antenatal and postnatal diagnosis have been reported from 23.8% to 50% for hydrocephalus and urinary tract anomalies (42.8%). Prenatal diagnosis has been found to be correct in about 60% of the urinary tract anomalies in another series. In the remainder the diagnosis was either incomplete or incorrect. Errors in the diagnosis resulted from difficulties in the differentiation of dilated ureter, intestinal dilation, or intraperitoneal cystic masses. On the contrary, many surgical anomalies are missed in spite of antenatal ultrasonography especially the gastrointestinal atresia, congenital diaphragmatic hernia and abdominal wall defects. With the expertise gained, the incidence has come down to about 10% at the tertiary care level institutes, for both false positive and the wrong diagnosis. Routine ultrasonographic screening during the antenatal period for congenital anomalies is far from satisfactory, especially in the peripheral areas in India from where cases are being referred to higher centers only at an advanced gestation age.

UNDERSTANDING THE NATURAL HISTORY OF SOME DISEASES

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 temporary physiologic impedance and the most do not require surgery. In a series of 197 newborns, ultrasonic follow-up showed that pyelocaliectasia resolved in 97%, only 3% presented with it in the postnatal ultrasound. Surgery was performed in 2% children.

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 hydronephrosis 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. 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 prenatal diagnosis of congenital lung malformations has been observed. Prenatal ultrasound appears reliable in the detection of pulmonary abnormalities but the variety of conditions identified postnatally suggests that specific prenatal diagnoses and prognosis should be avoided; prenatal counselling and perinatal management should be adapted accordingly. Antenatally diagnosed CDH turning out to be a CCAM is not uncommon.

The diagnosis of antenatally detected tumors (specially the neuroblastoma, rhabdomyosarcoma, teratoma) needs to be evaluated in the postnatal period for urinary catecholamines, growth in tumor size and the symptoms it produces. Aggressive therapy may be indicated in a few. Few cases may show complete resolution spontaneously. Follow-up however, is mandatory.

MATTERS OF CONCERN

1. The ISPAT (Indian Society for Prenatal Diagnosis and Therapy) conference at Delhi had very few members and participants from Paediatric Surgery. More number of Paediatric surgeons should participate in such conferences with full enthusiasm so that their role in early phase of decision-making is not diluted. However, we need to appreciate the global frontiers. The International society of fetal medicine and surgery (www.ifmss.org) holds an annual meeting with the participation of experts from various disciplines usually at a resort, this time it is being held in Island - Kona, Hawaii. This is truly an international platform in this field with an important role being played by paediatric surgeons.

2. Obstetricians, Radiologists and Geneticists should be encouraged to refer the patients with antenatally diagnosed congenital surgical anomalies in time for the paediatric surgeon’s opinion and discouraged from...
making their sole decision for termination.
3. The pioneers of fetal surgery meet yearly to discuss the relevance of fetal surgery today. The world leaders from their cumulative experience of the past 2-3 decades in the field are realizing that the indications for fetal surgery are more precise today and limited to a few anomalies. Fetal surgery has very little importance in developing countries where it is easier to reproduce another healthy baby rather than accepting a baby who is less than normal.
4. The government needs to be approached to accept and run programmes for promotion of the use of folic Acid and zinc therapy before and during pregnancy to prevent some of the well-known neural tube defects in at least 70-80% cases.

RECOMMENDATIONS

1. The paediatric surgeon has a very important role to play in the team for antenatal counselling of paediatric surgical anomalies.
2. 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 with the available facilities, helping them to decide the fate of the unborn child.
3. The decision should not be made on a single ultrasonographic scan and repeated evaluation is mandatory to filter out some of the false positive results, assure the parents to accept the defect and also reduce the diagnostic discrepancy reported.
4. An interdisciplinary approach is essential. The obstetrician should not take the sole responsibility of deciding the fate of the foetus with an antenatally diagnosed congenital surgical anomaly.

REFERENCES


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