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Antenatal diagnosis - ethics, challenges and reality: A pediatric surgeon perspective

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Abstract

Aims and Objective: To share the experience of pediatric surgeon while dealing with the birth defects in antenatal period with the changing law for medical termination of pregnancy in India.

Material and Methods: A retrospective observational study of 188 pregnant women presenting to the department of pediatric surgery for antenatal consultation. The women were followed up till the pregnancy was completed or terminated. Various parameters were noted down in the form of maternal age, educational status, gestational age, types of anomalies and organ system involved. There was change in medical termination of pregnancy (MTP) act which was relaxed in concern with gestational age.

Results: Mean gestational age (GA) of fetus was 27.26 weeks and 49 individuals had GA above 20 week. MTP rate in our study was 14.8%, where 6 terminations were considered unnecessary owing to increased anxiety. Anxiety level was high in 65 women. Educational status did not have a significant impact on the level of anxiety. Most common congenital anomaly was noted in genitourinary system. Most terminations and perinatal deaths were noted in Central nervous system (CNS) and cardiac system. More dilemma was involved in patients with suspected posterior urethral valve, congenital diaphragmatic hernia, CNS malformations.

Conclusion: Decision making in birth defects is a challenging task with the development in medical technologies. Most of them present after 20 weeks of gestational age. Relaxation of GA for MTP is going to be beneficial in dealing with serious anomalies. Couples should be given definitive plan of treatment after birth and sufficient time to answer their queries. In certain situations, the counseling should not be directive but should be supportive to help couple take a decision. Our decision should be bound with ethics, law and reasonable medical justification in helping parents to take the decision.

Keywords: Prenatal diagnoses, fetal diagnosis, fetal imaging, antenatal diagnoses, intrauterine diagnoses

Introduction

Paediatric surgeons are invariably involved while dealing with foetus having structural anomalies. Assurance to the parents on his ability to successfully treat the anomaly once baby is born is expected out of him. Many countries lack birth defect registry, but registers help in planning the health policies [1]. Pregnancy is very much a natural process; the advances in the technology is leading to significant medical interference. The concern has been raised by some nations as the medical termination of pregnancy (MTP) have raised drastically in recent decades [2]. First MTP was carried out in 1972 after the use of ultrasonography (USG) [3]. There are only few articles which gives paediatric surgeon's perspective to the antenatally detected anomalies [4, 5]. Here we have made an attempt to pen down the dilemmas, ethical issues, termination policies which could influence our decision making.

Material and methods

All the pregnant women who were referred to the department of pediatric surgery for antenatal consultation between August 2017 to January 2021 showing fetal structural abnormality in (USG) were included in the study. The initial consultation was done with the available USG findings. USG was repeated whenever necessary. Decisions were taken depending upon the existing pathology and gestational age (GA). A minimum of 30 minutes were spent with the expectant mother and her relatives (husband, father, mother or mother-in-law). In case of serious structural anomalies such as anencephaly, complex cardiac defects, renal agenesis the poor prognosis of the condition was explained and directed towards MTP. In case of correctable surgical anomalies, continuation of pregnancy was advised.

GA of twenty weeks was upper limit of termination of pregnancy as per our law at the time of collection of data. In special occasions i.e. Central nervous system (CNS) involvement, congenital airway abnormality etc fetal MRI was advised. Following parameters were recorded such as maternal age, maternal educational status, consanguinity in marriage, gestational age at presentation, any medical ailments in parents, family history of congenital or chromosomal abnormality, previous abortion, organ involved, assessment of anxiety level recorded using Hamilton anxiety rating scale (points >25 were considered to have moderate to severe anxiety) [6], counselling given and final outcome of the pregnancy were assessed. Our department was not involved in screening for aneuploidy. All the patients were screened in department of obstetrics before being referred to our unit.

Results

A total of 188 pregnant women were included in the study. The maternal age was ranging from 18 to 36 years (mean- 25.7 years). There were 24 women above 30 years of age. The gestation age at presentation varied from 14 to 37 weeks with a mean gestational age of 27.26 weeks. Forty-nine women presented to consultation before the GA of 20 weeks, 126 women presented after 20 weeks (no data regarding 13 patients). Sixty-nine were primigravida and 16 patients had gravida status more than 3. Thirty-eight women had history of one or more abortions. Deaths of their previous children were noted in 12 couples leading to 28 women without a living child (primigravida excluded). Consanguinity was noted in 38 couples. Genitourinary (GU) system was common organ system involved followed by nervous system, cardiovascular system and abdominal pathology. Eighteen patients had more than one anomaly. Six out of 16 cardiac anomalies had other associated congenital anomalies. Totally there were 63 pregnancy losses in the form of termination of pregnancy and perinatal deaths.

High levels of anxiety were noted in 65 mothers. Among them, 35 had no living children (including primigravida). The level of educational status did not have much bearing in the anxiety status. All but one had secondary schooling and 23 of these women had finished graduation. Anxiety in 33 mothers were justified as their fetuses had serious birth anomalies. Fifteen highly anxious mothers opted for termination. There were 20 perinatal deaths.

Total of 30 patients among 188, opted for termination of pregnancy. The gestational age varied from 14 weeks to 20 weeks. Among thirty women 16 were highly anxious. Multiple anomalies were noted in 7, urological anomalies were noted in 9, CNS involvement in 12, abdominal pathology in 1. Terminations in 5 women were considered unnecessary which included 4 cases of hydronephrosis and one with peritoneal calcification. Six terminations advised by us were under dilemma since outcome were not very sure, as in neural tube defects, Congenital Diaphragmatic hernia (CDH) and vertebral anomalies.

Among the 188 patients, we considered that 45 patients had life threatening anomalies such as neural tube defects, CDH, brain malformations, bladder outlet obstruction, congenital lung lesion, cardiac anomalies, renal agenesis, Tracheo-esophageal fistula (TEF), gross fetal hydrops etc. We had suggested termination in 15 patients. Majority of them had CNS involvement. There were 2 intrauterine deaths; one with cardiac defect and another with Arnold Chiari malformation. Perinatal death was noted in 14 of 45 fetuses. Two fetuses had CNS malformation, two had lung lesions, three had CDH, one patient

had meningomyelocele, two had suspected PUV, two had TEF with premature births, one patient had suspected tracheal stenosis and one patient had bilateral renal agenesis. All the above-mentioned fetuses were detected after 20 weeks hence we could not offer them pregnancy termination. Following four babies were healthy after birth and did not require any interventions. Foetuses with mass in mediastinum pushing the cardia, suspected TEF, common Atrio Ventricular canal with narrow pulmonary trunk and one had ventriculomegaly of 37 mm but didn't require any surgical intervention even after 6 months of follow up. Two children were taken against medical advice without undergoing surgery. One had tracheoesophageal fistula with neural tube defect another had neural tube defect with severe neurological deficit.

Total of 24 patients underwent surgical procedures after birth. Two patients died during follow up. Eleven had urological conditions, in which 8 had pelviureteric junction obstruction (PUJO) (one was operated after 1 year of follow up, one had bilateral PUJO operated on different occasion) and 3 patients had posterior urethral valve in which one died after 2 months due to renal failure. One patient with leaking meningomyelocele was operated (had neurogenic bladder). In abdominal pathologies, one patient had ovarian dermoid. Two patients with dilated bowel were noted to have ileal atresia and duodenal atresia respectively. Two patients had TEF, where one TEF patient was associated with duodenal atresia died after surgery. Two fetuses with cleft lip and palate were operated.

Thirteen patients had different postnatal findings compared to the USG findings in antenatal period. One of them was foetus-in-fetu which was reported as suspected intra-abdominal sacrococcygeal teratoma, one foetus with dilated bowel turned out to be intra-abdominal lymphatic malformation, one with enteric cyst turned out to be an ovarian dermoid. One foetus with peritoneal calcification, two fetuses with mega cisterna magna, one foetus with pleural effusion with ascites and one with distended bladder were found to have normal imaging studies after birth. Three patients had foetal MRI evaluation during the work-up. One for hydrocephalus along absent corpus callosum, another with shallow posterior cranial fossa and one of them with airway obstruction. Two patients had normal scan findings in 20 weeks scan but one was noted to have gross hydronephrosis (later diagnosed as PUV) and another had bilateral echogenic lungs in third trimester scan. All the above five patient did not survive.

We had two patients with infertility treatment, two patients with In-Vitro Fertilization (IVF), two patients with twin pregnancy (in both the cases only one twin was affected).

Discussion

Congenital abnormalities vary between 2-3.5% of total pregnancies where only 55% is detected prenatally and accounts for 30% perinatal mortality [7, 8, 9]. The antenatal counselling has ethical as well as legal aspects which needs consideration before making the decisions. It should also take care of anxiety and take away the guilt from the parents about the causation of the birth defects. Due to the availability of high-resolution ultrasonography, certain nonspecific findings are being identified such as increased nuchal fold thickness, hypoplastic or absent nasal bone, aberrant right subclavian artery, echogenic bowel, or short long bones, which otherwise wouldn't indicate a particular disease. We also had some other findings like enlarged cisterna magna, dilated rectosigmoid, and echogenic cardiac ventricle in our series. Initial recognition of such soft-markers were made in 1980s [10]. Importance of these, when

associated with any structural anomaly is unknown as parents also become more concerned with such findings. According to the American gynaecological and obstetrics society (AGOS) the overall termination rate has gone up to 50% for birth defects, among them 88% is for chromosomal abnormality and 32% for structural anomalies and are of the opinion that all the pregnant ladies should be offered aneuploidy screening and invasive testing^[11]. Lower gestational age, low educational status in mother, CNS lesions, chromosomal abnormality are some of the factors leading to low tolerance for termination. Seventy-five percent of the anomalies are detected in low-risk patients while screening. Ultrasonography has become an important part of pregnancy “work-up”. The success rate of detection of anomalies varies between 15-85%. The range is too wide as this depends on many variables such as expertise available, organ scanned, body habitus of pregnant lady and different centres. Specific scanning in tertiary setup has 2.7 times more yield compared to the peripheral hospital^[12, 13, 14]. Commonest anomalies to occur is urological abnormalities followed by neurological abnormalities. We also have same findings as in our study^[4, 11]. India has annual death rate of 606999 owing to congenital anomalies. Perinatal mortality was 5.5/1000 and neonatal mortality of 3.6/1000 in one of the studies. MTP was carried out in 7.5% according to one study, but in our case, it was 14.8%^[15]. In a study by Kumar and *et al.*, CNS involvement was more and GU system involvement was very low. The perinatal mortality was 8.8% in contrary to ours it was 14.3%. This could be considered limitation of our study due to referral bias as most of the serious anomalies were referred to us. Mean age of presentation their study was 31 weeks and 27% of their patients presented before 24 weeks of gestational age. In our study mean gestational age at presentation was 27.26 weeks with 26% of patients presenting before 20 weeks of gestation. About 80% world population is in developing countries, birth of disabled children is going to magnify the burden on these nations due to lack of facilities to cater differently abled ones. It may cause mental and physical taxing to the care givers. Authors in their study have opined that the medical terminations have become more safer and many parents and treating physicians are happy with more relaxed law to terminate the pregnancy for medical grounds and suggests that MTP should be allowed till term if there any serious abnormality in foetus^[16].

Law plays a very important role in making the policies related to these issues. It is based on the altitude of problem in their respective countries and willingness to spend on their population. In our country gender identification of foetus is an offence due to higher incidence of female foeticide and termination of the pregnancy after 20 weeks of gestation was prohibited, which has now been revised in May 2021. Hence it poses challenge for identification of certain gender influenced diseases, such as PUV. It also becomes difficult to follow up the diseases like congenital adrenal hyperplasia. In Netherlands routine screening of the pregnancy for any abnormality was not allowed until last 2-3 decades and were of the view that imparting lot of tests in pregnant women will be like considering pregnancy as disease process^[17]. India has witnessed 3 great amendments in MTP act in last 5 decades. One of the most sought, was allowing the termination of pregnancy up to 24 weeks and removal of upper time limit for MTP in case of severe birth anomalies after taking clearance from medical committee for gestation above 24 weeks. Seventy-four percent of our patients presented for consultation after 20 weeks^[18]. Decision of termination in anomalous foetus with uncertain prognosis (such as neural tube defects, congenital diaphragmatic hernia, posterior urethral valve) could be taken after following

them up for sufficient period of time. We did not have sufficient time with us before taking a decision for termination in about 6 of our patients. With the relaxed law for termination, 13 women in our study would have been benefited. At the same time the definition of severe congenital anomalies has not been made clear. Form the parents point of view all the anomalies in foetus seem to be grave for them. We noticed that 5 couple opted for termination with non-serious anomalies as they did not want a baby with even a minor defect. There is no clear-cut time indication or guidelines for advising termination. Most of the decision has to be individualised based on parents' decision, available resources and organ system involved. The decision making for the consultants are getting tougher as the increasing diagnostic options are available and it is highly sensitive to detect very small defects.

Since few decades the distinction between foetus and neonate are fading away. Presently medical technologies have been able to evaluate the foetus, provide intrauterine therapeutic care and has been able to manage viability issues of preterm babies giving an unborn child its own right in par with neonates and humanise the foetus. The fact that foetus can sense pain and already the mother is sensing a life within her, narrows down the distinction between the two, although both are still different in eyes of law, slowly this demarcation may also fade away. Hence people are of opinion that MTP needs to be restricted to the foetus suffering from severe form of disorders, as medical accidents can happen any time after the birth and we don't deprive them of the medical care when it occurs. Opponents of this, claim that only medical technology has made it possible to observe so much of detail about the foetus but in reality, it is not possible to humanise the foetus and decision about the continuation should be taken in best interest of the parents' patients^[19]. Woman right issue also needs to be addressed while making the decisions as she has to bear the brunt of carrying the malformed foetus and devote her time for the care of handicapped child in future. Many of the countries have relaxed the law in view of woman rights. We have provision for termination of pregnancy in our MTP act in case of contraception failures. In that case, it seems valid to demand for termination of pregnancy with foetal congenital defects irrespective of whether it is minor or major anomaly. First trimester foetal structural anomaly screening is now coming in becoming popular. It has been estimated that majority of the life-threatening and severe congenital anomalies can be detected in first trimester. Termination at this stage even before the expectant mother realises that there is a life within her, due to the absence of foetal movement may have less emotional disturbance^[20]. False positive rate is also considered to be low (0.09-0.6%) as compared to second trimester scan^[21]. We had four fetuses with serious anomalies on antenatal scanning, which escaped medical termination as they presented to us after 20 weeks but were doing well after birth. Foetal MRI has indication in CNS malformation or airway malformation which can change the line of management in 7-8% of cases^[22]. We had a primigravida young mother with airway obstruction as confirmed by foetal MRI. Continuing the pregnancy would mean, subjecting the young mother to risky EXIT procedure, led us to dilemma. Gestational age at consultation was 23 weeks and we had to continue the pregnancy which ended in neonatal death as they denied to undergo EXIT procedure.

MTP is acceptable strategy for reducing the burden of disability and genetic disorder in the society. Pregnant women consider USG and aneuploidy screen tests as an important part of the process and lack the purpose of these tests. Eventually pregnant women are not prepared to listen to bad news as they are not

prepared for the one. Pregnant ladies are also not aware of the available test to identify the anomalies [23, 24]. Population based government screening program is also not available in our country as noted in certain countries like Canada [25]. Government should look for prevention of these disease such as Neural tube defect. Vora et had determined the feasibility for fortification of tea with folic acid which may be helpful in reducing neural tube defects [26]. Anxiety level was assessed by Kemp *et al.* where they found that counselling by surgical colleagues reduced the anxiety in parents. Even without the counselling the anxiety tends to fall over a certain period. Lack of specific information and misinformation can increase the anxiety level. Early referral to the tertiary care centre for counselling may reduce the extended period of uncertainty [5]. Parents do seem to relate better if the surgeon comes with the definitive plan rather than being pragmatic yet entirely honest. Parents also expect sufficient time to ask questions, inform about the follow-ups and sympathy provided by the counselling person [26]. Still, we may come across difficult situations; such as recurrent pregnancy loses, child born by IVF, twin pregnancy where selective termination of affected twin is harmful to the normal foetus. This leaves us with the questions that whether couple should have an abnormal child after infertility treatment having been struggling for many years. In such situations the physician should explain about the pros and cons regarding the

continuation of pregnancy and if the parents wished to continue the pregnancy, counselling should not be directive but need to be supportive to the parents and give them the opinion regarding the treatment options available for the management of such children [27]. Two types of end of life decision have been proposed in MTP. One where straight away termination is carried out which may have long lasting consequences on parents another one is non-aggressive obstetric care where intervention needed to sustain life are foregone due to poor prognosis [28].

With the available technical advances, the task of decision making in pregnancy with deformity and suspected malformation is going to be difficult in coming. Pregnant woman should be given sufficient time to discuss with the obstetrician, pediatric surgeon or other specialist physicians (neurosurgeon, cardiologist), genesis/neonatologist and psychologist where counselling is done in a single chamber as a team. The counselling should be more supportive than divertive; family preference, women health and rights, best interest of unborn child needs to be addressed before taking a firm decision. Technology keeps advancing but our decision should be bound with ethics, law and reasonable medical justification in helping parents to take the decision. We have relaxation of law for MTP for severe congenital anomaly, but the tool has to use very wisely.

Table 1: Various parameters assessed in the study

Sl. No	Variables	Numbers	Percentage
1	Mean age of the mother	25.68 years	
2	Mean gestational age	27.26 weeks	
3	Age of presentation (among 175 women)		
	<20 weeks	28	16
	20-24 weeks	62	35.42
	>24 weeks	85	48.57
4	Gravida status (among 178 women)		
	Primigravida	69	38.76
	Gravida 2	58	32.58
	Gravida 3	38	21.34
	Gravida 4 or more	16	8.98
5	Abortion status		
	Once	25	13.29
	More than once	13	6.91
6	Death of previously born child	12	6.38
7	No living child (Other than primigravida)	28	14.89
8	Consanguinity		
	Second degree	32	17.02
	Third degree	6	3.19
9	Organ Involved		
	Genito urinary	96	51.06
	Nervous system	35	18.61
	Gastro Intestinal tract	16	8.52
	Limb and Skeletal muscle	6	3.19
	Cardiac vascular system	16	8.51
	Esophageal atresia and Tracheo-esophageal fistula	5	2.65
	Congenital diaphragmatic hernia	10	5.31
	Airway and thorax	7	3.72
	Cleft lip / cleft palate	3	1.59
	Hydrops fetalis	2	1.05
	Others	3	1.59
	Multiple anomalies	18	9.57
10	Intra uterine deaths	9	4.78
11	Perinatal deaths (Including still born)	26	13.82
12	Termination of pregnancy	30	15.95
13	High anxiety level in parents	65	34.57
14	Need for surgery after the baby is born	23	12.23
15	The postnatal diagnosis was not similar to antenatal findings	30	6.91

Table 2: Details of outcome in anxious women, foetus with lethal diseases, patients requiring the surgery and details of foetus being aborted

MA	GU	CNS	Abd	CVS	Tho	MSK	TEF	Others	MTP	PD	IUD	Surgical
1. Highly Anxious Mothers (65 women)												
8	34	10	2	4	6	3	4	3	15	17	3	8
2. Patients considered to be having lethal disease (45 patients)												
8	8	16	0	10	8	3	4	1	15	24	9	3
3. Patients undergoing surgery (23 children)												
2	11	1	4	none	1	none	1	2	-	2	-	23
4. Patient who opted for termination (30 patients)												
7	9	12	1	7	1	2	0	1	30	-	-	-

MA- Multiple anomalies, GU- Genito urinary, Neuro- Central nervous system, Abd- Abdominal pathology, CVS- Cardiovascular system anomalies, Tho- Thorax and congenital diaphragmatic hernia, MSK- musculoskeletal system including limbs and vertebra, TEF- trachea esophageal fistula, O- others, MTP- medical termination of pregnancy, PD- perinatal deaths- Neonatal and still birth, IUD- Intra uterine death, Surgical – patients who underwent surgery

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