MANAGEMENT OF ANTENATALLY DIAGNOSED BIRTH DEFECTS

Dr Sanjay along with senior obstetricians of this region is also actively involved in a project to find out the cost effective antenatal screening programme for diagnosing and managing birth defects in high risk pregnant women.

A healthy outcome of the pregnancy is a dream of every couple. The detection of any fetal anomaly can generate lot of stress and anxiety in the couple. Once a specific fetal anomaly is diagnosed, a detailed counseling has to be provided to the couple. The questions that the parents are supposed to ask are in relation to its management are:

- What is this particular anomaly?
- Is it minor or major or in other words correctible or correctible not?
- What is the best treatment available for this defect?
- Will this baby be perfectly alright after treatment?
- What are the risks, chances of survival and long term results of a surgical procedure required for the anomaly?
- Are the facilities for its treatment are available in this city or the baby has to be referred to some other place?
- What would be the approximate cost of treatment? etc.

The objective of counseling is to reduce the genetic burden as far as possible and to achieve the desired reproductive goal without undue harm to mother and fetus. This discussion during counseling should include: [1] the accuracy and limitations of the tests performed for diagnosing the particular anomaly and limitations and safety of other antenatal tests available or invasive procedures available to reconfirm the recently detected anomaly. [2] The best available antenatal and postnatal treatment for the diagnosed anomaly. What are the risks, chances of survival, expenses and long term results of a surgical procedure required for the anomaly?

The ultimate goal of their management is to improve the outcome of pregnancy that would indirectly reduce the neonatal mortality and morbidity due to birth defects. This goal can be achieved by preventing deliveries of babies having uncorrectable or incompatible anomalies and by providing better antenatal care and appropriate post natal surgical/medical treatment to the correctable anomalies. On the basis of this detailed information the couples are allowed to reach an independent and appropriate decision.

It is important that a pediatric surgeon be involved in discussing the postnatal management and counseling with the parents, as he is familiar with all the available options in the management of such babies. With the better understanding and advancements in the pediatric intensive care and anesthesia, most of the congenital malformations are correctible now. Babies suffering with such problems can now look forward to a normal life and often they do far better than their physicians expect initially.

THERAPEUTIC OPTIONS AVAILABLE FOR BIRTH DEFECTS DIAGNOSED ANTENATALLY

Broadly speaking there are two options:

[I]. MTP or abortion: Indicated for all anomalies that are uncorrectable or Incompatible with life or that would seriously affect the postnatal quality of life. Common among these are:

- Neural Tube Defects
- Anencephaly, Severe Hydrocephalus, Hydroencephaly
- Severe Chromosomal Abnormalities e.g. Down's Syndrome or Trisomy
- Bilateral renal agenesis & infantile polycystic kidney
- Major exomphalos, Cloacal extrophy
- Severe bone dysplasia e.g. Osteogenesis Imperfecta
- Multiple Anomalies occurring as a part of a Syndrome

If the tests show that fetus is abnormal and the parents elect to have the fetus aborted, most obstetricians prefer to terminate the pregnancy before 20 weeks

gestation, although up to 24 weeks is permissible by law in special circumstances. The treatment is not complete after an abortion as these cases would be referred for genetic counseling regarding the risk of recurrence in the next sibling and to take possible preventive measures if any [e.g. folic acid prophylaxis for NTDs].

[II]. To continue the pregnancy and manage the anomaly after delivery: This is indicated in cases:

[A] For all Correctable anomalies. In this condition a definitive surgical correction of the baby with a curative intention is done after delivery. Depending on the type of the anomaly, decisions are made regarding: 1. Does it need change in the time of delivery? 2. Does it need change in the mode of delivery [Caesar vs. normal]? 3. Does it need a maternal transfer to a better place near term? 4. Does it need any fetal intervention?

[B] Uncorrectable anomalies where mother refuses for MTP or if such anomaly is diagnosed beyond the safe limit of MTP then no option is left except to continue the pregnancy. Whatever management that we do is purely with the intention for palliation or temporary relief in the suffering of the baby till he survives. In these situations after delivery there is no scope for the definitive or curative treatment for the baby.

Factors determining the choice of treatment among the above two are:

1. Type of anomaly i.e Correctible or uncorrectable
2. Gestational Age at the time of detection
3. Associated Anomalies
4. Attitude of parents. That may further depend on their cultural, Social, financial background and also on the fact how many children they are already having or in other words how precious is the pregnancy for them. e.g. pregnancy occurring after many years or IVF, etc.
5. Available facilities to deal with such anomalies after delivery.

[B] FOETAL ANOMALIES BEST MANAGEGED AFTER TERM DILIVERY

Most correctable malformations that can be diagnosed in utero are best managed by appropriate surgical or medical therapy after delivery near term. This plan of management suits the following anomalies:

- Gut Atresia e.g. esophageal, duodenal, ileal, anorectal atresia
- Duplication cysts, Enteric cysts, Cholidochal cysts
- Small Omphalocele or exomphalos
- Unilateral multicystic or dysplastic kidney
- Unilateral Hydronephrosis
- Cystic Hygroma
- Cleft lip, palate

Some of the birth defects are serious or life threatening needing urgent operation while in others one can wait for some time till the baby becomes fit for surgery. Total correction of these malformations can be done in one stage in most of the cases. However, in some complex defects the total correction may take two or three stages.

Cesarean Section Vs Vaginal Delivery

A decision regarding a suitable mode of delivery would be taken for individual anomaly to see whether vaginal delivery is safe or it needs a Caesarian section. An elective Caesarian operation rather than a trial at vaginal delivery will be required for any fetal malformation that causes dystocia or there will be injury to the malformation during vaginal delivery or that will benefit from immediate surgical repair in a sterile environment. These are: hydrocephalus, large cyst, large hydronephrosis, Sacrococcygeal teratoma, big cystic hygroma, conjoint twins etc.

[C] FOETAL ANOMALIES REQUIRING PRETERM DELIVERY FOR EARLY CORRECTION EX UTERO

In cases where a continued gestation will have an adverse effect on a specific organ or system, a decision for early delivery would be taken. For example:

- Bilateral Hydronephrosis [PUJ obstruction]

- Posterior urethral valves
- Severe Hydrocephalus
- Diaphragmatic Hernia
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In such cases the risk of premature delivery will be weighed against risk of damage to the organ/system occurring by continuing the gestation. It is better to wait for at least 34 weeks till the lungst mature by surfactant production thus improving the survival of a premature baby by RDS. In such cases delivery would be planned and it will be ensured that facilities to deal with premature babies are available and also arrangements would be made for appropriate personnel to deal with such situation e.g. neonatologist, pediatric surgeon, anesthetist etc. If such facilities are not available in town or if parents wish for more advanced care for the baby, arrangements should be made for transfer of mother [i.e. fetus in situ] to a higher center for the delivery to take place.

[D] FOETAL ANOMALIES THAT NEED SURGICAL INTERVENTION BEFORE BIRTH

[Role of Fetal Surgery]

Yes, certainly intrauterine fetal surgery is a big hope for such babies. We can do a definitive correction of the anomaly in uterus. The purpose or idea is that if we correct the abnormality in uterus, normal development of the organ occurs later on and there is no further damage to organ. It is indicated for malformations that severely interfere with organ development and that, if alleviated, would allow normal fetal development to proceed. The indications are more or less the same as those for preterm delivery [c] with the difference that in preterm delivery we do a definitive correction after birth by managing it as a separate patient. There are only 3-4 dedicated centers allover the world where the facilities for fetal surgery are available. There is no such centre in India at present. Although a definitive correction has been performed successfully in fetus for some of the anomalies but that is confined mainly to experimental animals while in humans the total number of open foetal surgeries performed is still less than a few hundred throughout the world. The preferred and safer option is that in fetal intervention we keep ourselves confined to a minimal procedure or surgery that is required to alleviate or check the process.

There are mainly three approaches for the fetal intervention. First one is the open fetal surgery where we open the uterus, do the correction and close it to allow the pregnancy to continue e.g. in NTD coverage, open vesicostomy, lip repair, application of tracheal clip or repair for diaphragmatic hernia, large sacrococcygeal teratoma excision, open VP Shunt etc. Second one is called video endoscopic fetal surgery where the surgery is performed with the help of the endoscopes with minimal incision on uterus. This is more popular and safer option and can be used for all the conditions mentioned in the open fetal surgery. Thirdly, we can use ultrasound guided percutaneous route to do some temporary procedures like putting shunt for PUV, Hydrocephalus etc.

Moreover, the fetal surgery today is still not a very established entity. The enthusiasm of fetal surgery that was very high in 80s and 90s has come to a plateau. The reasons why it is still not very popular are: It needs a highly specialized and costly center. This not only need an ICU care for the mother but also need a fetal ICU. In spite of fetal ICU, there is very high fetal mortality and a significant rate of complications in mother also. Many times in spite of best efforts, you watch a fetus dying in front of you helplessly. It carries a significant risk to mother during pregnancy. Subsequently these mother would always need a Caesarian Section for delivery The reason being, at the time of mid gestation when we do fetal surgery, the lower segment of uterus is not formed so we have to go through mid uterus for fetal surgery. This increases the chances of rupture of uterus in Subsequent pregnancy. There is a very high incidence of premature labour as any stimulus to uterus like incision, endoscope or prick induces the labour.

The other important limitation of the fetal surgery is that during the pregnancy the correction can not be done at a time when the anomaly has really formed. In true sense it does not provide the definitive correction at a right time. At present the appropriate time for fetal surgery is around mid gestation i.e. around 18-20 weeks. Before that is not possible for the technical reasons. As we know that the formation of most of the major anomalies [e.g. NTD] are completed within 6- 8 weeks of gestation but we can diagnose it only around or after 12 weeks. Not only is this, even after their diagnosis, the surgery not possible at that time so we have to wait till 18-20 weeks. In other words a significant time elapses between the formations of anomaly till we do fetal surgery. So we cannot do much for the damage that occurs between their formation or

detection till they are corrected by fetal surgery. So the goal in fetal surgery is only partially achieved in that way. The fetal surgery will have best results only when the correction is done at a point of time when it is just formed and it is only possible when we can diagnose it at that time and simultaneously we are prepared to correct at that point of time which is not possible at present.