The Role of Radiographs in Fetal Autopsy

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The main objective of performing a fetal autopsy is to identify the etiology of fetal loss. This provides an explanation about the fetal loss to the members of the family and helps them adjust to their loss and cope with the unexpected bereavement. Requesting the family for fetal autopsy is an emotionally charged situation, as the fetal loss occurs in a setting where the family is looking forward to the birth of a normal neonate. The treating obstetrician with whom the family members have been associated, and have the utmost faith in, is the most appropriate person to obtain the consent for a detailed examination of the fetus. For this task, the obstetrician should know about the procedure and be informed about the utility of fetal autopsy to ascertain an etiology, which is necessary to counsel for recurrence risks and future reproductive options.

There are many publications supporting the utility of fetal autopsy in confirming/altering the antenatal diagnosis and helping in the counseling of the families for future reproductive options [1–3]. It thus serves as an audit for the services provided by the department of fetal medicine [4, 5]. In cases with malformations that do not conform to a syndrome, autopsy provides valuable information for the discovery of new syndromes and for defining their etio-pathogenesis [6].

In most publications from the West that examine the agreement between the observations at autopsy and antenatal diagnosis, the antenatal scans have been performed by persons with expertise in fetal ultrasounds. Vogt et al. [5] compared the results with previous studies and noted a statistically significant improvement in overall detection of

fetal anomalies on antenatal ultrasound. The improved technology and better diagnostic competence were the main reasons for this observation. India is a vast country and the current scenario here is different. Fetal medicine is a relatively new specialty and there are only a few specialized centers in the country. The quality of antenatal scans is therefore variable. This makes fetal autopsy valuable for making a correct diagnosis and counseling families as well as improving professional competence.

The standard protocol for fetal autopsy includes collecting information about the family, obstetrical and medical history, details of imaging, results of screening for aneuploidy, and invasive testing, if performed, and other investigations that have been carried out. As Desilets et al. [7] state “observe everything from head to toe including dysmorphism, malformations, deformations,” and photograph anything “out of the normal.” Weigh and measure everything that can be weighed and measured, and compare with normal values. Internal examination is performed as per standard procedure [8, 9], with macroscopic and histology evaluation. Special tests such as for storage disorders, infections, and hematologic disorders are carried out as indicated.

Radiographs form an important part of fetal autopsy and some advocate it in every fetal death [10], whereas others use it only in cases of nonchromosomal fetal anomalies [6]. One of the very early studies in India by Pahi et al. [11] concluded that a revised diagnosis by fetal autopsy changed recurrence risk in 29.5 % of cases. In 12 of 21 cases where there was a change in diagnosis after fetal autopsy, a noninvasive external examination and radiographs were useful.

There are many situations where a standard fetal autopsy cannot be performed. It may be due to the family not consenting for autopsy, the clinician not being able to

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convince the family about the utility of the full autopsy [12] or the expertise for an autopsy not being available locally. The declining rates of fetal autopsy in India as well as the rest of the world [13], necessitate the identification of protocols for noninvasive and minimally invasive fetal autopsy. Wright et al. [10] reviewed the options available for investigating perinatal deaths when families decline consent for the standard autopsy. They concluded that valuable information can be obtained from external examination, radiographs and placental and cord evaluation in most cases, with a limited number requiring MRI and tissue biopsies. Radiographs were recommended in all cases to identify skeletal and soft tissue abnormalities. Radiographs are also useful to determine fetal gestational age [14, 15].

The paper by Deka et al. [16] in this issue of the journal, describes how plain, noninvasive radiographs can provide useful information to counsel families for future pregnancies, if full autopsy cannot be done. They discuss the utility of radiographs in the presence of fetal congenital malformations and correlate it with the gold standard of fetal autopsy. They successfully identified skeletal dysplasias, congenital diaphragmatic hernia and neural tube defects. However, hydrocephalus and nonsyndromic arthrogryposis would be better delineated by a good external examination conducted by a dysmorphologist rather than by radiographs. The best noninvasive modality for delineating CNS malformations such as hydrocephalus, is fetal postmortem MRI.

It is well recognized that radiology is the single most important investigation in a fetal skeletal dysplasia. Deka et al. [16] report a change in counseling based on the revision of the diagnosis of the type of skeletal dysplasia after a radiographic study. We have also reported before that postnatal external examination and detailed radiographic assessment of the fetus, especially of the pelvis, limbs, skull and spine, are essential to identify the type of skeletal dysplasia [17].

In a recent study, Breeze et al. [18] compared conventional autopsy to postmortem MRI and external examination with additional investigations like placental histology, skeletal radiographs and cytogenetics. They concluded that the results obtained were equivalent with both techniques, but more information was obtained with imaging guided biopsies. Thayyil et al. [19] have recently reported a concordance of 99.4 % between conventional and minimally invasive autopsy and conclude that where appropriate expertise is present, it could be an acceptable alternative. The postnatal fetal MRI as well as specific organ biopsies can be of tremendous help in decision making for the bereaved families, especially where conventional autopsy is not acceptable. To their clear message that “at least obtain a full body radiograph in the presence of a skeletal

abnormality in the fetus” we would like to add “MRI if possible for internal structures and save umbilical cord or some soft tissue like spleen for DNA extraction” for molecular studies later on.

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