



Perinatal Pathology and Fetal Medicine

Raj P. Kapur¹

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Fetal medicine is a multidisciplinary field in which data collected from multiple sources to formulate the best care plan possible. This issue of the *Journal of Fetal Medicine (JFM)* highlights perinatal pathology, a core component in any well-designed fetal medicine program. Gross and microscopic studies of the placenta and embryo, fetus, or neonate can provide invaluable insights into the etiology and/or pathogenesis of a wide range of conditions with important implications, particularly those related to genetic counseling and the management of future pregnancies. The practice of perinatal pathology requires a thorough understanding of normal developmental anatomy, ability to recognize and document deviations from the norm, knowledge of common congenital disorders, and skilled utilization of other informational and/or laboratory resources to investigate rare conditions. Proficiency in this subspecialty requires years of practice and continued education with regard to newly defined syndromes, advances in molecular genetics, and human embryology.

Perinatal pathology is ideally practiced by a pathologist specifically trained to examine products of conception. However, in many communities, by necessity, the role is filled by a geneticist, obstetrician, or other professional, whose formal training may not have included pathology per se. The articles in this issue are written by experts in the field of perinatal pathology to explicitly review some fundamental practice guidelines and illustrate basic principles that guide the subspecialty. Topics were chosen with other members of the fetal medicine team in mind,

including nonexperts who may be called upon to perform some type of perinatal pathology examination. Collectively, these papers provide a good overview of how to approach different aspects of the exam, the type of information that can be collected with systematic analyses, and how this data can be integrated with other laboratory studies and correlated with clinical findings.

Examination of the placenta is a cornerstone of perinatal pathology, especially when a pregnancy is complicated by multiple gestation, premature delivery, intrauterine growth restriction, infection, or fetal demise. In his article on “Placental Gross Examination”, Dr. Sunil Jaiman describes a logical step-by-step approach to examination of this organ, explaining how gross findings are used to direct microscopic evaluation. Dr. Jaiman emphasizes findings that may explain fetal demise. A common cause of fetal demise and other adverse pregnancy outcome is fetal thrombotic vasculopathy (FTV), which is the subject of the scholarly contribution by Drs. Marsden and Comstock. Readers learn how FTV is recognized and the challenges associated with distinguishing antemortem FTV from nonspecific postmortem histopathological vascular changes. They summarize conflicting published data regarding the relevance of hereditary thrombophilia to the pathogenesis of FTV and the role for a thrombophilia workup in affected pregnancies.

Placental mesenchymal dysplasia is less common than FTV and less well known to many clinicians, but has garnered increased attention recently because of its sonographic confusion with molar pregnancy, clinical associations with assisted reproduction technology, and Beckwith–Wiedemann syndrome, and fascinating genetic findings. Dr. Linda Ernst’s comprehensive review of placental mesenchymal dysplasia (PMD) discusses all of these aspects. Readers will finish this paper prepared to consider

✉ Raj P. Kapur
raj.kapur@seattlechildrens.org

¹ Department of Laboratories, OC.8.720, Seattle Children’s Hospital, 4800 Sand Point Way NE, Seattle, WA 98105, USA

PMD in the appropriate clinical contexts and able to distinguish PMD from “look-alikes” using a combination of clinical, pathological, and molecular data.

Evaluation of the placenta is only one-half of the perinatal pathology equation. Examination of the baby, especially a dysmorphic fetus or infant, poses another set of challenges, which benefit from a systematic approach. Dr. Sunil Jaiman, Dr. Ratna Puri, and Lance Erickson provide just such a framework in their articles on “Fetal Examination”, “Fetal Dysmorphology”, and “Examination of the Fetal Heart”, respectively. Dr. Puri explains how to approach the fetal exam and illustrates how information should be collected and integrated from many sources. She introduces many of the basic terms used to describe fetal anomalies, their correct usage, and how they reflect contemporary understanding of pathogenesis or etiology. Her focus repeatedly returns to the clinical value of information obtained from the fetal examination, with many specific examples of dysmorphic findings that directly impact genetic counseling and management of subsequent pregnancies. Dr. Jaiman builds on this foundation and takes us through the basic steps of a fetal autopsy, including external and internal gross examination. He describes how clinical findings guide the dissection approach (e.g., posterior fossa dissection to exclude suspected Dandy–Walker malformation) and how gross impressions affect sampling for histology.

Examination of the fetal heart can be especially difficult, given that at mid-gestation, the organ is less than a centimeter in greatest dimension. Correct detailed descriptions of cardiac anatomy are paramount, as specific anomalies

have different prognostic, surgical, and genetic significance. Correlation with prenatal sonographic diagnosis is critical, in part, to insure that the latter is accurate. The dissection method advanced by Lance Erickson captures the key cardiac findings through a logical sequence of in situ manipulations, which readers should be able to apply immediately and master with practice.

Nonimmune fetal hydrops is one of the most common phenotypes encountered in fetal medicine. Many affected fetuses die and are referred to the pathologist, who faces a lengthy differential of diagnostic possibilities. Dr. W. Tony Parks has faced this situation many times. In this issue of *JFM*, Dr. Parks describes fetal hydrops, mechanisms thought to lead to fluid accumulation in the fetus, and a logical means to narrow the diagnostic possibilities. He stresses the value of considering pathogenesis as a basis for grouping possible specific etiologies, an approach that guides observation and reduces the likelihood that a key diagnostic feature is overlooked.

A common theme in each of the articles in this issue is the importance of good communication between pathologist and clinician, a philosophy which is appropriately embraced by the *JFM*. If such communication is to succeed, it is essential that all caregivers have some appreciation for how and why products of conception are evaluated. Hopefully, content related to perinatal pathology will remain a vital part of *JFM* and readers can look forward to more information on this important topic in the future.