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EDITORIAL COMMENTARY

Lessons from an Autopsy Study of Fetal Renal Malformations

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Since time immemorial, autopsy studies have laid the foundation of modern medicine, enabling us to challenge established facts and providing a tour into the human anatomy [1]. In the current era of high-resolution imaging, anatomy comes alive in details hitherto unimaginable. Anatomical studies of the kidney and urinary tract are particularly useful to pediatric nephrologists, and fetal anatomy provides useful insights into disease pathology.

In this issue of the Journal, Ganesan and colleagues have reported anomalies of the kidney and urinary tract from 45 fetal autopsies; and correlated the findings with those on antenatal ultrasonography [2]. They have presented a spectrum of abnormalities in the number, size, position, and shape of the kidney(s), cystic kidney diseases, and lower urinary tract abnormalities. The majority of fetuses had syndromic or multisystem anomalies associated with congenital anomalies of the kidney and the urinary tract (CAKUT). Hydronephrosis was the most common abnormality, noted in 40% of fetuses.

The authors have reported that 30% of all abnormalities comprised renal cystic disease. This unexpectedly high proportion has been explained by the fact that severe cystic dysplasia, when present bilaterally, is incompatible with life. The reader should remember that autopsy findings do not reflect the distribution of CAKUT in childhood, since severe fetal anomalies are commonly fatal and minor findings are often missed in antenatal ultrasonography as well as in asymptomatic children. Therefore, the high prevalence of 7% for autosomal recessive polycystic kidney disease in this autopsy study is not comparable to any childhood cohort studied till date [3]. Similarly, since the majority of fetal hydronephrosis resolves spontaneously, the prevalence of postnatal hydronephrosis is considerably lower.

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There are other differences between this study and similar past publications. For example, previous studies reported extrarenal anomalies in nearly 90% of fetuses with renal anomalies [4, 5]. The diagnosis of well-described syndromes has implications for prenatal testing in subsequent gestations. Fetal autopsy provides the opportunity for genetic diagnosis, which can further improve outcomes in subsequent pregnancies.

A noteworthy finding is a discordance between findings on ultrasonography and autopsy in nearly 40% of cases. The discrepancy between antenatal ultrasound and autopsy findings varied across settings and was as low as 10%–14% in previous studies from developed countries [5, 6]. This discordance reiterates the importance of expertise in fetal radiology to improve the precision of fetal diagnosis, which has important implications for fetal and postnatal outcomes. However, diagnosis of specific abnormalities can inadvertently disclose gender, which treads a sensitive domain with implications for both medicine and law. Magnetic resonance imaging has emerged as an alternative to fetal autopsy, given its good sensitivity, and can be used to conduct virtual autopsies [7].

In summary, the fetal autopsy studies on CAKUT provide valuable insights into renal and genitourinary tract development and pathobiology, which can improve the outcomes of the next pregnancy. Genetic diagnosis from autopsies and virtual autopsies may pave further paths in this area. Indeed, the dead do teach the living.

Declarations

Conflict of Interest None.

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