A Pathologists’ Assistant’s Critical Role in the Proper Diagnosis of Gastroschisis and Trisomy 18

Abstract:
Although gastrochisis is a frequently studied abdominal wall defect, it continues to be researched as many believe the condition has an environmental component. Unlike omphalocele, a similar abdominal wall defect, the organs are not covered by a protective sac. Thus, the severity of gastrochisis depends on the number of protruding organs as well as the duration they are exposed for. In this particular case, a woman from Brazil presented to a hospital in Pennsylvania for a follow-up on the condition of her fetus. Through techniques such as gross and microscopic dissection, as well as microarray testing, her fetus was discovered to have gastrochisis and trisomy 18.

Background:
Gastrochisis is a rare type of abdominal wall defect that occurs during gestation. The term originates from the Greek word laprosthesia, which means bellied.1 In this condition, the abdominal muscles do not form properly, creating a hole adjacent to the umbilicus. Further, the baby’s intestines and other organs may protrude outside of the body and come into contact with amniotic fluid. Over time, the abnormal condition can lead to irritation and damage. Since its discovery, gastrochisis has been linked with young maternal age. While the exact cause is unknown, some theories include a single or a combination of environmental factors. Through early screenings and other tests, gastrochisis can be diagnosed during pregnancy or after birth. Yet, one of the most concerning aspects of the condition is its increasing worldwide incidence.

This case involves a 40-year-old woman G1, PO, who presented to the hospital at 32 weeks and 5 days gestation. She had previously received prenatal care in Brazil where she was told that her fetus had congenital anomalies not compatible with life outside of the womb. Thus, the patient had been planning on having a palliative labor and delivery. However, upon presentation to the hospital in Pennsylvania, the patient was found to have no fetal heart tones. She also developed an abdominal defect. However, unlike omphalocele, the organs are not covered by a protective sac. Although gastrochisis may be associated with genetic aberrations, they are more commonly seen as late onset in older women. The most important distinction is the presence or absence of an outpouching of peritoneum, or a sac. Although gastrochisis may be associated with genetic aberrations, they are more commonly seen in cases of omphalocele. Thus, the particular case involving the mother from Brazil is unique, as her fetus was found to have Trisomy 18 on microarray testing.

Incidence:
Many studies have been performed in order to evaluate the prevalence of gastrochisis on a state or national level. For instance, in Texas, researchers discovered an incidence of 5.13 cases per 10,000 births.2 The most comprehensive study of gastrochisis to date took place in two parts within the United States from 1995 to 2005, and 2006 to 2012. Performed by Jones et al.3,4, births in 14 states, the equivalent of 29 percent of all births within the United States, were evaluated in order to assess the prevalence of gastrochisis. Researchers discovered that the incidence of gastrochisis increased by thirty percent between the two time periods, from 4,369 cases per 12,014,244 births to 4,497 cases per 9,264,540 births.5 However, as more studies were performed throughout the world, an apparent discrepancy in incidence was discovered. This led researchers to believe that gastrochisis has strong environmental causes, due to the stark contrast in frequency between countries. Interestingly, in addition to regional disparities, incidence also appears to vary in accordance with race. In a multi-state study by Abby M. Jones et al., gastrochisis was present at much higher rates in non-Hispanic black mothers compared to non-Hispanic white mothers.6 This same population also had the greatest increase in prevalence from 1995 to 2012, with a growth of 263%.7 However, there has been controversy over the misdiagnosis and reporting of the prevalence of gastrochisis. Depending on the location, gastrochisis with Trisomy 18 may only be recorded as Trisomy 18, and therefore not included in the gastrochisis count.8 It follows that areas recording both anomalies would have higher recorded rates of gastrochisis, than those accounting only for the chromosomal abnormality. Thus, it is imperative that gastrochisis be recorded separately from other abdominal wall defects and chromosomal aberrations in order to have a more accurate representation of its incidence.

Methodology:
A bedside ultrasound was performed and intrauterine fetal demise was confirmed. Before discharging the patient, plans for an autopsy and chromosome analysis with micro-array testing were made.