Gastroschisis Associated with Lower Limb and Spinal Congenital Anomalies

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ABSTRACT
Gastroschisis is not a very rare congenital deformity, but extragastrointestinal association is rare, if any present, in that condition, an alternative diagnosis should be considered, like Pentalogy of Cantrell, Limb-body wall complex, etc. Other birth defects are always associated with gastroschisis, most commonly, abnormalities of the cardiac and genitourinary. The present case is one of the gastroschisis to highlight the associations of spinal and lower limbs anomalies, with two-vessel short umbilical cord and severe oligohydramnios in primiparas.

Key words:
Gastroschisis, limb-body wall complex, omphalocele

INTRODUCTION
Gastroschisis is a right-sided, small, and full-thickness paraumbilical defect of the abdominal wall that occurs in 1 of 4000 births.[1] Unlike an omphalocele, the herniated bowel is in direct contact with amniotic fluid. Theories concerning the etiology of gastroschisis are usually considered to be the result of a vascular insult.[2]

Cardiac and genitourinary abnormalities have been associated[3-8] with gastroschisis, but presence of extra-gastrointestinal anomalies warrants search for alternative diagnosis. In the presented case, gastroschisis associated with spinal and lower limb anomalies in early age group mother (primiparous) has been presented.

CASE REPORT
A 22-years-old full-term primiparous patient underwent an ultrasound examination, we found large paraumbilical defect with herniation of stomach, small and large bowel loops, liver, gallbladder, and right kidney. Herniated stomach and bowel loops were rest over internal Os [Figure 1]. Amount of liquor was very less and fetus looked flexed and twisted in breech presentation. Spinal anomalies were observed block cervical vertebrae with spina bifida of cervical and lumbar spines. In lower limbs anomalies, mildly short, thin left femur and left tibia with very thin fiber-like left fibula were seen. Both feet were rudimentary, especially left foot, with no defined toe while right foot was with four abnormal toes. Left iliac bone was rudimentary. Umbilical cord was short and had two vessels [Figure 2].

After delivery, we took photographs of dead abnormal newborn. We found that newborn has large anterior abdominal paraumbilical defect. Stomach, intestinal loops, liver, gallbladder, and right kidney were herniated through the large abdominal defect [Figure 3].

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Figure 1: USG image of fetal abdomen showing herniated bowel loop, right kidney, and fetal liver resting on maternal internal Os
lower limbs were mildly short; left leg being thinner with deformed feet and small three buds of toes while right foot presented with four abnormal toes [Figure 4]. Left side hip was small with kypho-scoliotic curvature of vertebral column. Rudimentary male genital organs were seen. Umbilical cord was short and had two vessels. Head, face, neck, thorax, and both upper limbs were normal.

CT scan and radiograph of newborn were also obtained [Figures 5-7] and the above said findings were corroborated by the images.
DISCUSSION

Gastroschisis is a congenital anterior abdominal wall defect, adjacent and usually to the right of the umbilical cord insertion. Gastroschisis has no covering sac and no associated syndromes. This differentiates it from an omphalocele, which usually is covered by a membranous sac and more frequently is associated with other structural and chromosomal anomalies. In addition, however, gastroschisis may be associated with gastrointestinal anomalies such as intestinal atresia, stenosis, and malrotation.[6]

By the study of Stoll C et al. of Omphalocele and gastroschisis and associated malformations, they assessed these associated malformations ascertained between 1979 and 2003 in 334 262 consecutive births. Of the 86 patients with omphalocele, 64 (74.4%) had associated malformations. These included patients with chromosomal abnormalities (25, 29.0%) and non-chromosomal syndromes. Malformations of the musculoskeletal system (31, 23.5%), urogenital system (27, 20.4%), cardiovascular system (20, 15.1%), and central nervous system (12, 9.1%) were the most common, other congenital malformations in patients with omphalocele and non-syndromic multiple congenital anomalies (MCA). Of the 60 patients with gastroschisis, 10 (16.6%) had associated malformations.[10]

Mastroiacovo P et al. performed an international study to identify malformation patterns and to evaluate the role of maternal age in non-isolated cases of gastroschisis and associated defects. Case-by-case information from 24 registries, all members of the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR) were evaluated. Their results showed that of 3 322 total cases, 469 non-isolated cases were registered (14.1%): 41 chromosomal syndromes, 24 other syndromes, and 404 MCA. Among MCA, four groups of anomalies were most frequent: CNS (4.5%), cardiovascular (2.5%), limb (2.2%), and kidney anomalies (1.9%). No similar patterns emerged except two patterns resembling limb-body wall complex and omphalocele-extrophy-imperforate anus-spinal defects (OEIS). In both of them, the gastroschisis could be misclassified. Chromosomal trisomies and possibly non-syndromic MCA are associated with an older maternal age more than isolated cases. On consideration of their data and the most valid studies published in the literature, the best estimate of the proportion of gastroschisis associated with major unrelated defects is about 10%, with a few cases associated to recognizable syndromes. Recognized syndromes with gastroschisis seem to be so exceptional that the well documented and validated cases are worth being published as interesting case report. An appropriate case definition in etiological studies should include only isolated gastroschisis after an appropriate definition of isolated and non-isolated cases and a thorough case-by-case review.[11]

Singal R et al. reported a rare case of a newborn baby with an abdominal wall defect, together with multiple congenital abnormalities and diagnosed as gastroschisis. There were multiple defects seen as spinal deformity, imperforate anus, esophageal fistula, and lower limb deformity (congenital talipes equinovarus) along with the webbing of neck. There were also ischemic changes present over the left upper limb in the form of cyanosis. The diagnosis made was gastroschisis and Omphalocele along with spinal deformity.[10] In the presented case, head, face, neck, thorax, and both upper limbs were normal.

CONCLUSION

Although Gastroschisis has rare associated malformations, but if present, Pentalogy of Cantrell, Limb-bodywallcomplex, etc., should be considered as an alternative diagnosis. Gastroschisis may be associated with gastrointestinal anomalies. Other birth defects are associated with gastroschisis, most commonly, abnormalities of the cardiac and genitourinary. The present case was not completely considered within any known alternative diagnosis of Gastroschisis-associated complex, non-syndromic and syndromic anomalies. The presented case of gastroschisis, therefore, highlight the associations of both spinal and lower limbs anomalies in primiparous, which was proven to be a rare case.

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