DOI: 10.5958/j.2319-5886.3.2.101

International Journal of Medical Research & Health Sciences

www.ijmrhs.com Volume 3 Issue 2 (April - Jun) Coden: IJMRHS Copyright @2014 ISSN: 2319-5886

Received: 29th Jan 2014 Revised: 28th Feb 2014 Accepted: 12th Mar 2014

Case report

GASTROSCHISIS: RECENT TRENDS, EMBRYOLOGY, MATERNAL AND INFANT RISK FACTORS

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ABSTRACT

The term gastroschisis is derived from the Greek word *laproschisis*, meaning "bellycleft." It was used in the 19th and early 20th centuries by teratologists to designate all abdominal wall defects. Gastroschisis occurs in approximately 1 in 2,300 live births, and mortality for gastroschisis may approach 10 percent. Several maternal risk factors suspected to be associated with gastroschisis have been investigated, and there is a consistent association with young maternal age. The association of low birth weight also could be attributed to the fact that data demonstrate that the birth weights of babies with gastroschisis are significantly lower than those of the general population and are similar in different populations. Recently, Stevenson et al. proposed that gastroschisis is caused by the failure of the sac and yolk duct, as well as of the vitelline vessels, to initially incorporate to the allantois and later to the body stem. The specific role of the genetic component in the etiology of gastroschisis is unclear. Although there are reports of familial cases, gastroschisis occurs mostly as a sporadic event. It was observed that 4.7% of cases have at least one affected relative and the risk of recurrence is 3.5% among siblings. Definitive treatment is surgical. The timing and technique for surgical closure depends on the degree of intestinal inflammation, size of the defect and the newborn's general condition. In general, the prognosis is good with a survival ≥90%, but in developing countries the risk of death may be as high as 60%.

Keywords: Gastroschisis, Yolk duct, Vitelline vessels

INTRODUCTION

The term gastroschisis is derived from the Greek word *laproschisis*, meaning "bellycleft." It was used in the 19th and early 20th centuries by teratologists to designate all abdominal wall defects.

Gastroschisis is a congenital defect of the abdominal wall in which the baby's intestines, and sometimes other abdominal organs, protrude from the belly through a small hole. Gastroschisis occurs in approximately 1 in 2,300 live births[1], and mortality for gastroschisis may approach 10 percent. Rates of gastroschisis have been increasing in many developed and developing countries with no tenable explanation or specific known causes for this trend. In 1994, the prevalence rate was 2.78 per 10,000 live births, and increased to 3.54 per 10,000 live births in 2011. Rates of gastroschisis are particularly high among younger mothers, including teen moms.

In most cases gastroschisis is an isolated birth defect; however, it also can be associated with other defects, particularly those with possible vascular mechanisms of origin. Reported overall incidence of concurrent anomalies is 7 to 30 percent, which may include anencephaly, cleft lip and palate, ectopia cordis, atrial septal defect, diaphragmatic hernia, scoliosis, syndactyly and amniotic band syndrome. Although

these anomalies are not related directly to the gastroschisis defect, they contribute to the morbidity.[2]

CASE REPORT

A term male baby was delivered by emergency cesarean section to a 24yr old primigravida mother due to fetal distress. Mother did not had regular antenatal check ups and was not a registered case. Birth weight was 2.6 kgs. Baby had severe respiratory distress at the time of birth and gradually deteriorated and did not respond to the resuscitatory measures and succumbed about one hour after birth (figure -1). On examination baby had right sided anterior abdominal wall defect measuring about 3.5 x 4 cm. There was evisceration of stomach, small and large bowel loops which were thickened and edematous. No membrane was covering the eviscerated bowel loops. The umbilical cord had two arteries and one vein. No other obvious external congenital anomalies were detected.

Fig1: Male baby born with gastroschisis.

Table 1: Risk factors associated with Gastroschisis

Parental occupation (eg, printer/computer manufacturing factories)
Young maternal age
Hispanic race
Poor maternal education
Low socioeconomic status
Lack of prenatal care
Nulliparity
More than one elective abortion
Short interval between menarche and first Pregnancy
Chorionic villus sampling
Residence surrounding landfill sites
Maternal diet (low alpha-carotene, low total glutathione, high nitrosamines)
Low pregnancy body mass index

Table 2: Potential Teratogens associated with Gastroschisis

Organic chemicals/solvents
Cyclooxygenase inhibitors (Aspirin, Ibuprofen)
Decongestants
Acetaminophen
Oral contraceptives
Maternal smoking
Alcohol
Illicit drugs (cocaine, amphetamine)
X-ray irradiation in early pregnancy

DISCUSSION

Several maternal risk factors (Table -1 & 2) suspected to be associated with gastroschisis have been investigated, and there is a consistent association with young maternal age (< 20 years of age). Torfs et al. reported a tenfold increased risk of gastroschisis occurrence where maternal age was between 15 and 19 years of age. Body mass index (BMI) and nutrient deficiencies in maternal dietary intake also are being considered as possible risk factors for gastroschisis.[3] Low alpha-carotene and low total glutathione and high nitrosamine intake during the trimester prior to conception have been associated with gastroschisis. This led to the hypothesis that younger age of mothers may lead to maternal fetal competition for nutrients with the result being maternal dietary inadequacy. [3, 4] Lam et al. found a higher risk of gastroschisis for underweight mothers and a lower risk for overweight mothers, and the California Birth Defects Monitoring Program revealed that underweight young mothers who presented with a Body Mass Index less than 18.1 had a greater risk of having a child with gastroschisis.[5] The association of low birth weight also could be attributed to the fact that data demonstrate that the birth weights of babies with gastroschisis are significantly lower than those of the general population and are similar in different populations. These findings support the notion that a normally functioning intestinal tract is essential for normal fetal growth.[6]

Table 3: Theories Regarding Embryogenesis of Gastroschisis

Author	Theory
Duhamel[7] (1963)	Teratogenic insult resulting in defective differentiation of the somatopleural mesenchyme
Shaw [8] (1975)	Rupture of a hernia of the umbilical cord at the site of involution of the right umbilical vein
DeVries [9] (1980)	Abnormal right umbilical vein atrophy resulting in weakness and defect of abdominal wall, with failure of epidermal differentiation
Van Allen[10] (1981)	Vascular disruption theory
Hoyme[11] (1983)	Omphalomesenteric artery insult with disruption of umbilical ring

Over the years, various authors proposed different hypotheses for the development of gastroschisis (Table -3). Recently, Stevenson et al. proposed that gastroschisis is caused by the failure of the sac and yolk duct, as well as of the vitelline vessels, to initially incorporate to the allantois and later to the body stem. It has been determined that there is a second perforation in the abdominal wall, as well as that of the umbilical ring, through which the midpoint of the intestine (Meckel point) is connected to the externalized vitelline structures. These are attached to the bowel abnormally, separating it from the body stem, which causes a failure in the incorporation of the umbilical stalk. As a result, the gut is extruded into the amniotic cavity without remnants of yolk sac or amnion so that the midpoint of the intestine is always externalized and there is an absence of vitelline remnants in the umbilical cord.[12] The location on the right of the defect can be explained by the tendency of the yolk stalk to move to this side due to the presence of the heart and more rapid growth of the left lateral wall.[12, 13.]

The specific role of the genetic component in the etiology of gastroschisis is unclear. Although there are reports of familial cases, gastroschisis occurs mostly as a sporadic event. It was observed that 4.7% of cases have at least one affected relative and the risk of recurrence is 3.5% among siblings.[14] Gastroschisis is usually detected by ultrasound after 18 weeks gestation because before week 14, the process of physiological herniation of the mid-intestine has not been completed.[15-18] Measurement of AFP (α-fetoprotein) in maternal serum between 16-18 weeks of gestation is useful for the detection of abdominal wall defects and the acetylcholinesterase/ pseudocholinesterase index to distinguish wall defects such as gastroschisis with neural tube defects.[14] When maternal alpha-fetoprotein (AFP) levels are elevated, obstetricians look for defects by having the expectant mother undergo a detailed prenatal ultrasound. With gastroschisis, this test will show loops of bowel (intestines) floating freely in amniotic fluid. More frequent ultrasounds are generally recommended to continue monitoring the fetus.

Multidisciplinary pre and postnatal management is required. Controversy remains today regarding the timing and route by which delivery should be performed. It is known that elective termination via cesarean section after 36-37 weeks gestation and before the onset of labor prevents passage through the birth canal, which decreases the risk of contamination with bacterial flora and mechanical damage in the viscera. However, a significant difference has not been shown in terms of complications or survival.[19]

Definitive treatment is surgical. The timing and technique for surgical closure depends on the degree of intestinal inflammation, size of the defect and the newborn's general condition.[20-23]

Primary surgical closure within 24 hours after birth is preferred, but if there is viscero-abdominal disproportion (present in 20-49% of cases), gradual reduction with silo is necessary to avoid complications. Surgical repair should be performed between 6 and 10 days of extrauterine life. In general, the prognosis is good with a survival ≥90%, but in developing countries the risk of death may be as high as 50-60%. The leading causes of mortality are related to prematurity, neonatal sepsis, intestinal complications related to intestinal ischemia, acute renal failure or multiple organ failure.[24]

CONCLUSION

Epidemiologic studies from the United States and other developed countries around the globe have reported an increased prevalence of gastroschisis over

a wide geographic distribution. Although environmental and maternal factors have been suspected, the cause of gastroschisis remains unclear, and no single cause has yet been implicated. Universally, there is a significant association of gastroschisis with young maternal age along with smoking, leading to speculations of a teratogen related to modern lifestyle that remains to be identified. Also, it is possible that gastroschisis may be related to a combination of factors working synergistically, rather than an isolated single event or exposure. This rising prevalence of gastroschisis has been described as an epidemic, emphasizing the importance of continued monitoring and evaluation of patho-genetic factors. The potential association of gastroschisis with medications, diet, and other maternal factors could have implications for pregnancy planning similar to neural tube defects. Thus, it is an important public health issue, highlighting the need for a more complete multicenter epidemiologic study.

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