



GASTROSCHISIS-A CASE PRESENTATION

Dr Ajith Kumar¹, Dr Roja VR^{2*}

¹Consultant Radiologist, Malabar Hospitals and Urology Centre. & Baby Memorial Hospital Calicut, Kerala

²Research Scientist (M)-I, ICMR-NTF HI Project, ENT and HNS Department, AIIMS Raipur

Conflicts of Interest: Nil

Corresponding author: Dr Roja VR

Abstract:

Introduction: Gastroschisis is a congenital anomaly, where the evisceration of abdominal contents through the defect in the anterior abdominal wall. Over 90% case can be detected in antenatal ultrasound. The present case was reported in Malabar Multi speciality hospital and urology center Calicut, Kerala, during a foetal ultrasound scan in the second trimester.

Case presentation: 29-year-old primigravida learned that her unborn child was detected with gastroschisis, during 16th-18th week of pregnancy and subsequent ultrasound examination also detected as complete gastroschisis.

Discussion and conclusion: The exact cause of gastroschisis is not clear, since it is a multifactorial disease.

Key words: GS, USG, AWD, Exomphalos, Omphalocele

INTRODUCTION

The term gastroschisis derived from Greek for stomach cleft or fissure, leading others to suggest the technically more correct term Laproschisis⁽¹⁾, it was used in English literature by Calder in 1733, first case had been described as early as 1557 by Lycosthenes⁽²⁾. Typically reviewed in conjunction with exomphalos, omphalocele and prune belly syndrome, still the aetiology of gastroschisis is stand alone, predisposing risk factors are different, clinical management and associated malformations⁽³⁻⁷⁾. Gastroschisis differs from other anterior abdominal wall defects, here bowel herniates without a covering membrane, through a defect adjacent to umbilicus, nearby and always right. Certain cases though rarely escape from such ready classification, similarities between exomphalos and gastroschisis exists⁽⁸⁻¹¹⁾. The incidence of its detection has been dramatically increased in the last century, probably due to the wide spread use of diagnostic imaging modalities particularly USG, for antenatal scans, 1980-90^(1,2,4,5).

The incidence AWD (abdominal wall defects) were 1 in 3659 live births over this 4-year period, with gastroschisis occurrence 18%. A detailed review of incidence of both Gastroschisis and Exomphalos reported to the National Congenital Malformation Notification scheme in England and Wales between 187 and 1993, found that increased in incidence of GS over first five years of study, with highest incidence were 1.35/10000 live births, in 1991⁽²¹⁾. Further studies conducted by Rankin et al⁽²²⁾, in North of

England, the year 1986 and 1996 found that rising incidence of GS rising to 4.72/10000 live births in 1996.

Similar study in North Carolina, USA identified the peak incidence of GS, it was documented that 4.46/10000 live births, in 2000⁽²⁵⁾. One European study conducted in between 1980-84 and 1995-2002⁽²⁴⁾, showed nearly a fourfold rise of GS. Geographical variation was again noted, with a peak incidence of 4.48/10000 live births in Mainz Germany compared with 0.31 in Tuscany (Italy)⁽²⁴⁾. Generally, the incidence was higher in Northern European countries compared to those bordering the Mediterranean⁽²⁴⁾.

Most recognised risk factors include, young maternal age^(1,21,22,26-28). Loane et al found a seven-fold increase in the relative risk of GS in mothers under the age of 20, compared to other age groups. Young maternal age linked to other cofactors include cigarette smoking, use of recreational drugs, low socio-economic status, poor nutritional status, young age at the time of pregnancy and previous terminations^(1,3,29-33). Relatively low incidence of other associated anomalies, despite with GS, appears increasing evidence of a genetic contribution with recent identification of specific gene polymorphism⁽³²⁾.

Increasing evidence, from twin studies, animal models and epidemiological data, represents GS is a true malformation rather than a disruption which has

occurred following normal development (26,38). Feldkamp et al (26), discredited traditional explanations, suggest that GS may result from herniation of bowel into the amniotic cavity, through a lateral ventral wall defect, resulting from failure in development of a body wall fold. The classical right sided presentation anomaly, explained by the relative positions of umbilical cord and yolk sac, where the latter generally to the right, supporting their hypothesis (4,5,26). Development of GS malformation occurs very early in gestation, between third and fifth post conceptual weeks (26), since many would be unaware of pregnancy and optimal pre and peri conceptual care is highly limited (26,31,33).

Case presentation:

29-year-old primigravida, married for the last ten years. learned that her unborn child was detected with gastrochisis, during 16th -18th week of pregnancy and subsequent ultrasound examination also detected as complete gastrochisis. A defect of 32mm in the anterior abdominal wall and herniation of small bowel loop detected, right side of the umbilical cord. It was not covered by membrane and the bowel wall oedema is quite obvious. Foetus floats in sufficient liquor volume. No other anomalies detected.

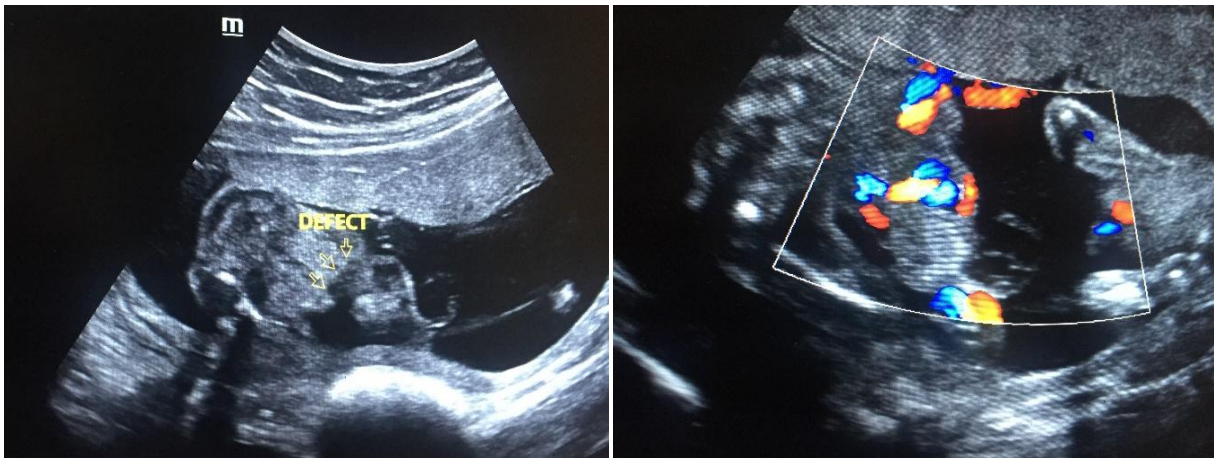


Figure 1: and 2: shows the defect in the anterior abdominal wall, with evisceration of the small bowel. The umbilical cord is also seen.

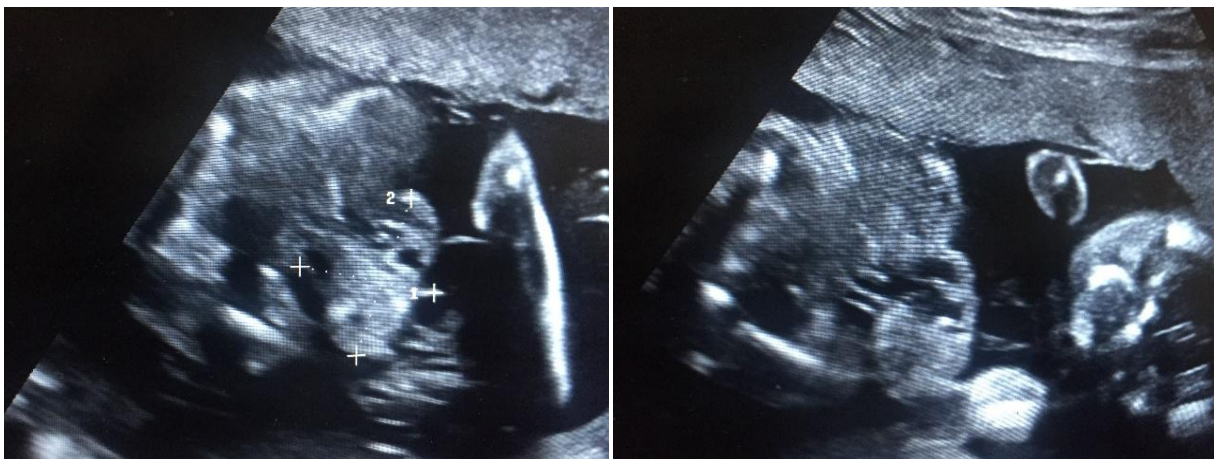


Figure 3 and 4: The eviscerated small bowel shows wall oedema; vascularity could not be ascertained in the wall.

The case presented in Malabar Institute of Research Centre, Calicut, Kerala, India. The case referred from Obstetrics and gynaecology department to Radiology department for a early detailed anomaly scan during 16th week and case diagnosed with Gastrochisis. Later with the request and consent of mother and father along with family members decided to terminate the pregnancy during 18th week.



Figure 5: shows after the expulsion of the foetus, shows herniated and oedematous bowel.

Discussion:

Increased prenatal surveillance needed in a diagnosed case of gastroschisis and delivery could be relocated in a tertiary obstetric hospital. Serial surveillance of fetal parameters needed with US, indicated in view of intrauterine growth retardation, still birth and premature delivery^(3,47,53). A western Australian study maintained with 122 cases of GS, followed up with serial US evaluation, assessment of amniotic fluid volume and bi weekly fetal heart rate monitoring from 32weeks onwards⁽⁵⁰⁾. Towers et al⁽⁵⁴⁾ suggested antenatal monitoring from 32weeks onwards, while David et al⁽³⁾ advocated fetal monitoring every 2weeks from diagnosis. Prematurity occurs in upto 60%,with 10 and 31%having an associated birth defect^(47,55).Relatively common associations include gastrointestinal atresia,⁽⁵⁶⁾ and undescended testis^(3,57,58).IUGR affected population or class of fetus with GS, affected between 30 and 70% foetuses^(53,59).the cause of IUGR is hypothesised ,either due to inadequate supply of nutrients or due to protein loss from exposed viscera.

The exposed bowel is vulnerable to injury which can range in severity from volvulus with loss of the entire midgut to a more localised intestinal atresia or stenosis, to widespread inflammatory peel or serositis. It makes bowel loops indistinguishable from

one another^(56,58). Inflammatory develops after 30 weeks of gestation and its aetiological factors include bowel wall exposure to amniotic fluid and intestinal lymphatic obstruction⁽⁶⁰⁾. Most devastating complication of GS,is unpredictable fetal death which usually occurs in third trimester^(60,64).caused due to acute midgut volvulus or acute compromised blood flow to the eviscerated bowel^(62,65,66). Recent evidence suggests a generalised cytokine mediated inflammatory response ensues, and this may explain how the conventional fetal surveillance methods fail to prevent still birth^(49,50,66,67). overall still birth rates are 10%, there appears to be an association with still birth and abnormal amniotic fluid volume (3,50). Reid et al⁽⁵⁰⁾ reported 50% of still birth occurrence in oligohydramnios compared to 16.7% with high amniotic fluid volume. Furthermore 70% Of pregnancies with abnormal amniotic fluid volume were delivered preterm compared with 30% cases where the amniotic fluid volume was normal⁽⁵⁰⁾. Degree of confidence in detecting GS via sonography is 75% and for omphalocele it is 77.3%.

Differential diagnosis: Omphalocele, bladder exstrophy, body stalk anomaly and periumbilical blood clots.

Summary: GS particularly affects young mothers of low socio-economic status and other risk factors including cigarette smoking and usage of illicit drugs. Its incidence reported to be 4.72/10000 live births. With the advent of sonological evaluation in early pregnancy with subsequent serial monitoring would help to diagnose and further management of the cases. Thwarting the complications and reduce the incidence of IUGR is the major challenge for the obstetrician.

Consent: Written informed consent was obtained from the patient for publication of this case report and accompanying images.

References:

1. Wilson RD, Johnson MP (2004) Congenital abdominal wall defects: an update. *Fetal Diagn Ther* 19:385–398
2. Bernstein P (1940) Gastroschisis, a rare teratological condition in the newborn. *Arch Pediatr* 57:505–513
3. David AL, Tan A, Curry J (2008) Gastroschisis: sonographic diagnosis, associations, management and outcome. *Prenat Diagn* 28:633–644
4. Islam S (2008) Clinical care outcomes in abdominal wall defects. *Curr Opin Pediatr* 20:305–310
5. Jacob C, Langer M (2003) Abdominal wall defects. *World J Surg* 27:117–124
6. Maksoud-Filho JG, Tannuri U, da Silva MM et al (2006) The outcome of newborns with abdominal wall defects according to the method of abdominal closure: the experience of a single center. *Pediatr Surg Int* 22:503–507
7. Lee TC, Barshes NR, Nguyen L et al (2005) Gastroschisis and biliary atresia in a neonate: uncommon presentation or common precipitant. *Eur J Pediatr Surg* 15:434–436
8. Gow KW, Bhatia A, Saad DF et al (2006) Left-sided gastroschisis. *Am Surg* 72:637–640
9. Suver D, Lee SL, Shekherdimian S et al (2008) Left-sided gastroschisis: higher incidence of extraintestinal congenital anomalies. *Am J Surg* 195:663–666
10. Chen CP (2007) Ruptured omphalocele with extracorporeal intestines mimicking gastroschisis in a fetus with Turner syndrome. *Prenat Diagn* 27:1067–1068
11. Weber TR, Au-Flieger M, Downard CD et al (2002) Abdominal wall defects. *Curr Opin Pediatr* 14:491–497
12. Kiesewitter WB (1957) Gastroschisis: report of a case. *AMA Arch Surg* 75:28–30
13. Bianchi A, Dickson AP (1998) Elective delayed reduction and no anesthesia: ‘minimal intervention management’ for gastroschisis. *J Pediatr Surg* 33:1338–1340
14. Schlatter M, Norris K, Uitvlugt N et al (2003) Improved outcomes in the treatment of gastroschisis using a preformed silo and delayed repair approach. *J Pediatr Surg* 38:459–464
15. Goulet O, Sauvat F (2006) Short bowel syndrome and intestinal transplantation in children. *Curr Opin Clin Nutr Metab Care* 9:304–313
16. Zaccara B, Iacobelli A, Calzolari A et al (2003) Cardiopulmonary performances in young children and adolescents born with large abdominal wall defects. *J Paediatr Surg* 38:478–481
17. Moore TC (1963) Gastroschisis with antenatal evisceration of intestines and urinary bladder. *Ann Surg* 158:263–269
18. Mann L, Ferguson-Smith MA, Desai M et al (1984) Prenatal assessment of anterior abdominal wall defects and their prognosis. *Prenat Diagn* 4:427–435
19. Lafferty PM, Emmerson AJ, Fleming PJ et al (1989) Anterior abdominal wall defects. *Arch Dis Child* 64:1029–1031
20. Moore TC (1977) Gastroschisis and omphalocele: clinical differences. *Surgery* 82:561–568
21. Tan KH, Kilby MD, Whittle MJ et al (1996) Congenital anterior abdominal wall defects in England and Wales 1987–93: retrospective analysis of OPCS data. *Br Med J* 313:903–906
22. Rankin J, Dillon E, Wright C (1999) Congenital anterior abdominal wall defects in the north of England, 1986–1996: occurrence and outcome. *Prenat Diagn* 19:662–668
23. Rankin J, Pattenden S, Abramsky L et al (2005) Prevalence of congenital anomalies in five British regions, 1991–99. *Arch Dis Child Fetal Neonatal Ed* 90: F374–F379
24. Loane M, Dolk H, Bradbury I (2007) Increasing prevalence of gastroschisis in Europe 1980–2002: a phenomenon restricted to younger mothers? *Paediatr Perinat Epidemiol* 21:363–369
25. Laughon M, Meyer R, Bose C et al (2003) Rising birth prevalence of gastroschisis. *J Perinatol* 23:291–293
26. Feldkamp ML, Carey JC, Sadler TW (2007) Development of gastroschisis: review of hypotheses, a novel hypothesis, and implications for research. *Am J Med Genet* 143A:639–652
27. Forrester MB, Merz RD (2006) Comparison of trends in gastroschisis and prenatal illicit drug use rates. *J Toxicol Environ Health* 69:1253–1259
28. environmental risk factors for gastroschisis and omphalocele in the National Birth Defects Prevention Study. *J Pediatr Surg* 44:1546–1551
29. Houglund KT, Hanna AM, Meyers R et al (2005) Increasing prevalence of gastroschisis in Utah. *J Pediatr Surg* 40:535–540
30. Lam PK, Torfs CP (2006) Interaction between maternal smoking and malnutrition in infant risk of gastroschisis. *Birth Defects Res Clin Mol Teratol* 76:182–186
31. Siega-Riz AM, Olshan AF, Werler MM et al (2006) Fat intake and the risk of gastroschisis. *Birth Defects Res Clin Mol Teratol* 76:241–245

32. Torfs CP, Christianson RE, Iovannisci DM et al (2006) Selected gene polymorphisms and their interaction with maternal smoking, as risk factors for gastroschisis. *Birth Defects Res Clin Mol Teratol* 76:723–730
33. Werler MM (2006) Teratogen update: pseudoephedrine. *Birth Defects Res Clin Mol Teratol* 76:445–452 876 *Pediatr Surg Int* (2010) 26:871–878 123
34. Werler MM, Mitchell AA, Moore CA et al (2009) Is there epidemiologic evidence to support vascular disruption as a pathogenesis of gastroschisis? *Am J Med Genet* 149A:1399–1406
35. Werler MM, Sheehan JE, Mitchell AA (2002) Maternal medication use and risks of gastroschisis and small intestinal atresia. *Am J Epidemiol* 155:26–31
36. Fielder HM, Poon-King CM, Palmer SR et al (2000) Assessment of impact on health of residents living near the Nant-y-Gwyddon landfill site: retrospective analysis. *Br Med J* 320:19–22
37. Hwang P-J, Kousseff BG (2004) Omphalocele and gastroschisis: an 18 year review study. *Genet Med* 6:232–236
38. Vermeij-Keers C, Hartwig NG, van der Werff JF (1996) Embryonic development of the ventral body wall and its congenital malformations. *Semin Pediatr Surg* 5:82–89
39. Murphy FL, Mazlan TA, Tarheen F et al (2007) Gastroschisis and exomphalos in Ireland 1998–2004. Does antenatal diagnosis impact on outcome? *Pediatr Surg Int* 23:1059–1063
40. Vegunta RK, Wallace LJ, Leonardi MR et al (2005) Perinatal management of gastroschisis: analysis of a newly established clinical pathway. *J Pediatr Surg* 40:528–534
41. Drewett M, Michailidis GD, Burge D (2006) The perinatal management of gastroschisis. *Early Hum Dev* 82:305–312
42. Richmond S, Atkins J (2005) A population-based study of the prenatal diagnosis of congenital malformation over 16 years. *Br J Obstet Gynaecol* 112:1349–1357
43. Algert CS, Bowen JR, Hadfield RM et al (2008) Birth at hospitals with co-located paediatric units for infants with correctable birth defects. *Aust N Z J Obstet Gynaecol* 48:273–279
44. Fillingham A, Rankin J (2008) Prevalence, prenatal diagnosis and survival of gastroschisis. *Prenat Diagn* 28:1232–1237
45. Davis RP, Treadwell MC, Drongowski RA et al (2009) Risk stratification in gastroschisis: can prenatal evaluation or early postnatal factors predict outcome? *Pediatr Surg Int* 25:319–325
46. Palomaki GE, Hill LE, Knight GJ et al (1988) Second-trimester maternal serum alpha-fetoprotein levels in pregnancies associated with gastroschisis and omphalocele. *Obstet Gynecol* 71:906–909
47. Nicholas SS, Stamilio DM, Dicke JM et al (2009) Predicting adverse neonatal outcomes in fetuses with abdominal wall defects using prenatal risk factors. *Am J Obstet Gynecol* 201:383–386
48. Cedergren M, Selbing A (2006) Detection of fetal structural abnormalities by an 11–14-week ultrasound dating scan in an unselected Swedish population. *Acta Obstet Gynecol Scand* 85:912–915
49. Luton D, Guibourdenche J, Vuillard E et al (2003) Prenatal management of gastroschisis: the place of the amnioexchange procedure. *Clin Perinatol* 30:551–572
50. Reid KP, Dickinson JE, Doherty DA (2003) The epidemiologic incidence of congenital gastroschisis in Western Australia. *Am J Obstet Gynecol* 189:764–768
51. Srivastava V, Mandhan P, Pringle K et al (2009) Rising incidence of gastroschisis and exomphalos in New Zealand. *J Pediatr Surg* 44:551–555 52.
52. Kandasamy Y, Whitehall J, Gill A et al (2010) Surgical management of gastroschisis in North Queensland from 1988 to 2007. *J Paediatr Child Health* 46(1–2):40–44
53. Puligandla PS, Janvier A, Flageole H et al (2004) The significance of intrauterine growth restriction is different from prematurity for the outcome of infants with gastroschisis. *J Pediatr Surg* 39:1200–1204
54. Towers CV, Carr MH (2008) Antenatal fetal surveillance in pregnancies complicated by fetal gastroschisis. *Am J Obstet Gynecol* 198:686–695
55. Payne NR, Pfliegerhaer K, Assel B et al (2009) Predicting the outcome of newborns with gastroschisis. *J Pediatr Surg* 44:918–923
56. Fleet MS, de La Hunt MN (2000) Intestinal atresia with gastroschisis: a selective approach to management. *J Pediatr Surg* 35:1323–1325
57. Lawson A, de La Hunt MN (2001) Gastroschisis and undescended testis. *J Pediatr Surg* 36:366–367
58. Lao OB, Larison C, Garrison MM et al (2010) Outcomes in neonates with gastroschisis in U.S. children's hospitals. *Am J Perinatol* 27:97–101
59. Franchi-Teixeira AR, Weber Guimaraes BM, Nogueira B et al (2005) Amniotic fluid and intrauterine growth restriction in a gastroschisis fetal rat model. *Fetal Diagn Ther* 20:494–497
60. Japaraj RP, Hockey R, Chan FY (2003) Gastroschisis: can prenatal sonography predict neonatal outcome? *Ultrasound Obstet Gynecol* 21:329–333
61. Marder AL, Moise K Jr, Chuang A et al (2008) Amnioexchange for the treatment of gastroschisis—an in vitro study to determine the volume and number of exchanges needed. *Fetal Diagn Ther* 23:95–99
62. Kohl T, Tchatcheva K, Stressig R et al (2009) Is there a therapeutic role for fetoscopic surgery in the prenatal treatment of gastroschisis? A feasibility study in sheep. *Surg Endosc* 23:1499–1505

63. Fasching G, Haeusler M, Mayr J et al (2005) Can levels of interleukins and matrix metalloproteinases in the amniotic fluid predict postnatal bowel function in fetuses with gastroschisis? *J Pediatr Surg* 40:1887–1891
64. Cohen-Overbeek TE, Hatzmann TR, Steegers EA et al (2008) The outcome of gastroschisis after a prenatal diagnosis or a diagnosis only at birth. Recommendations for prenatal surveillance. *Eur J Obstet Gynecol Reprod Biol* 139:21–27
65. Ledbetter DJ (2006) Gastroschisis and omphalocele. *Surg Clin North Am* 86:249–260
66. Chabra S (2006) Management of gastroschisis: prenatal, perinatal and neonatal. *Neoreviews* 7:e419–e427