FETAL ABDOMINAL WALL AND GASTROINTESTINAL ANOMALIES

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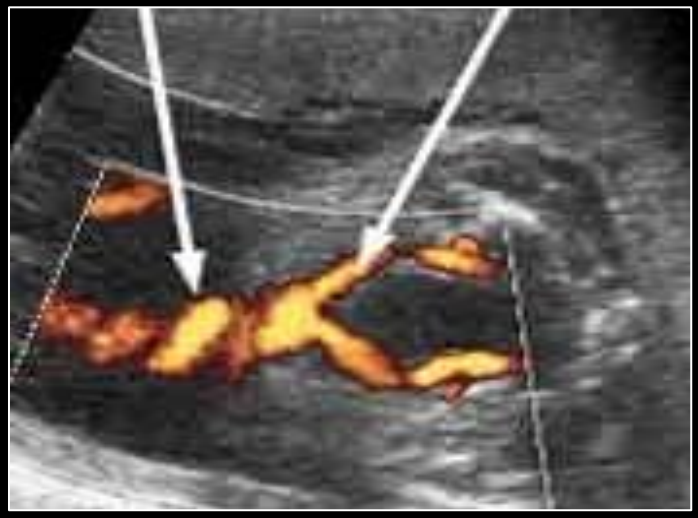
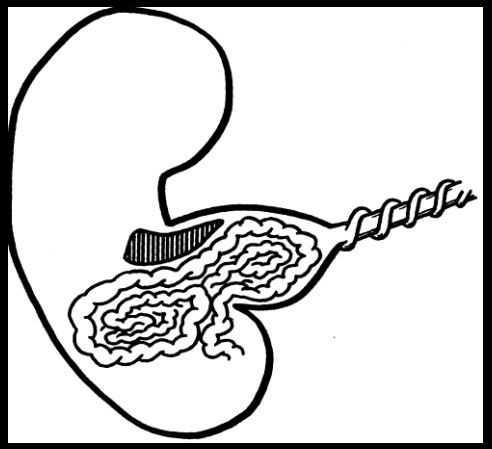
Cerrahpaşa Tıp Fakültesi Kadın Hastalıkları ve Doğum Anabilim Dalı

Abdominal Wall



Omphalocele

- 1/3000- 5000 pregnancy
- Skin,facia, muscles absent
- Covered by peritoneum and amnion (translucuent membrane)
- Umbilical cord at the top of sac

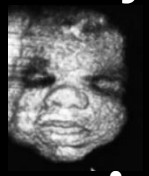


- Associated Anomalies **(50-80 %)**

Cardiac	8%-52%
Renal and external genitalia	2%-45%
Limb	2%-30%
Ectopia cordis	2%-8%
Caudal fold defect	2%-8%
Central nervous system	2%-30%
Gastrointestinal tract	28%-79%

- **Chromosomal Abnormality (30-50 %)**
  - **Trisomy 21,13,18, Triploidy, Turner**

- **Syndromes**

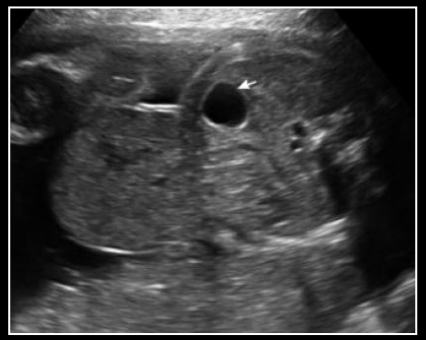


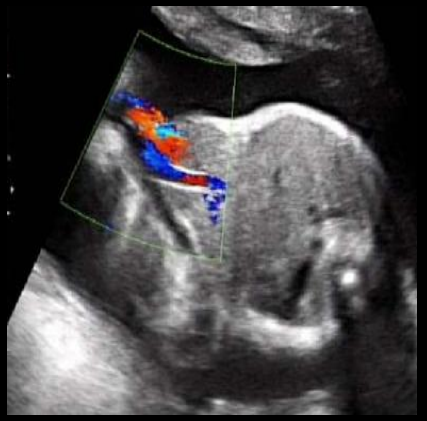
  - **Beckwith – Wiedemann (4-19 %)**
  - **Cantrell Pentology**
  - **Shprintzen**
  - **Carpenter**
  - **Limb-body wall complex**
  - **OESİS (**Omphalocele, blader exstrophy, imperforate anus, spinal defects)

- **İsolated**
  - **%10-30 not isolated**

Prenatal Diagnosis

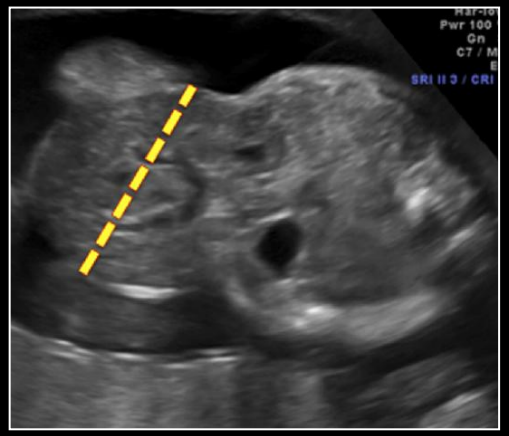
- Easy
- 11-14 GW / 70-100 %
  - 9-11 gw / physiological herniation
    - CRL>44mm
    - Size>7mm
    - Liver herniation
  - <12-14 gw pren. diagn?
- 18-24 GW / 80-100 %

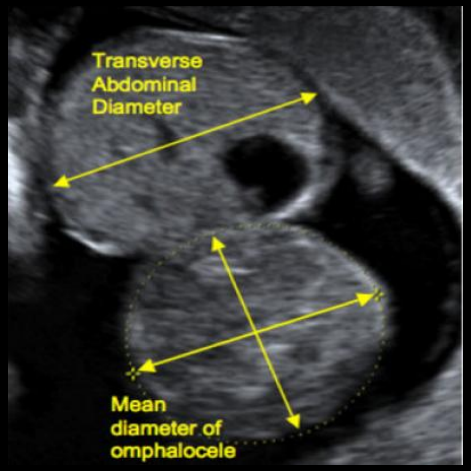




Prenatal Management

- İn the sac
  - Small hernia
  - İntestines
  - Liver
- Measurement
  - Giant Omphalocele / >4-5 cm or >75% liver





- Karyotype
- Detailed USG
- Fetal Eko

- Prognosis
  - İsolated 90 – 96%
  - Associated ↓
- Defect size / Content
  - ? (OD/AC, OC/AC, OD/AD)

- Abortus- IUD 5-10%
- Preterm Deliv. 25-30%
- FGR 15-39%

Should We Measure Fetal Omphalocele Diameter for Prediction of Perinatal Outcome?

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Fetal Diagn Ther 2014;35:44–50

Table 4. Summary of studies that have examined antenatally the association between fetal omphalocele size and adverse perinatal outcome

Study	Omphalocele study group	GA diagnosis, weeks	Omphalocele ultrasound size or criteria	Comments
Hughes et al. [19]	n = 22 isolated: 7 major anomalies: 10 minor anomalies: 2 chromosomal defects: 3	21.3 (12–40)	OD/AD >0.6	no association with postnatal mortality
Kamata et al. [11]	n = 31 isolated: 10 associated anomalies: 21	29.9±4.9	small, no liver (n = 12) giant, liver herniation (n = 12) ruptured (n = 7)	1 fetal death; 11 primary closures with 2 deaths 2 deaths before surgery; 10 staged closures with 2 deaths 1 fetal death; 1 death before surgery; 5 staged closures with 4 deaths
Biard et al. [12]	n = 8 isolated or minor anomalies	18.5 (12–24)	giant omphalocele (more than 75% of liver in the sac)	all cases without primary closure need for mechanical ventilation; time to first oral feed; duration of parenteral feeding
Nicholas et al. [16]	n = 33 associated anomalies: 15	18.8	liver herniation	death, hospital stay, >2 surgeries, need of supplemental feed at the time of hospital discharge, sepsis: OR = 7 (1.2–41.7)
Patel et al. [10]	n = 28 associated anomalies: 17	≤20	minor, <5 cm (n = 14) major, >5 cm (n = 11) giant, entire liver in the sac (n = 3)	all: isolated, primary closure, survived all survived: 4 primary closures, 6 secondary closures, 1 conservative management all silo closures due to ruptured omphalocele, death

Study	Omphalocele study group	GA diagnosis, weeks	Omphalocele ultrasound size or criteria	Comments
Montero et al. [13]	n = 25 major anomalies: 4 minor anomalies: 6	25.0±5.7	OD/AC >0.24 OD/HC >0.21	primary closure not feasible: OR = 7 (1.1–41.3) primary closure not feasible: OR = 7.7 (1.3–45) NICU stay, need for mechanical ventilation, time to first oral feed
Kleinrouweler et al. [7]	n = 45 isolated: 18 associated anomalies: 12 chromosomal defects: 15	13+5 (10+3 to 35+5)	OC/AC >0.62 (n = 2) OC/AC >0.66 (n = 15) OC/AC >0.57–0.75 (n = 13)	for herniation of the liver: sensitivity 87%, specificity 90% respiratory insufficiency: sensitivity 67%, specificity 78% staged surgery: sensitivity 100%, specificity 100%
Tassin et al. [14]	isolated: 54	12 (11–14)	OD/AD >0.8	hospitalization in the intensive care unit for more than 42 days; need for respiratory assistance and/or need for parenteral feeding >21 days
Present study	n = 47 isolated: 38 minor anomalies: 9	26.6 (16.6–35.8)	OD/AC ≥0.26	postnatal death: LR = 4 (1.9–7.5) need for intubation on the first day: LR = 2.6 (1.2–5.7) need for two-step surgery or mesh: LR = 5.9 (1.9–14.4)

- **Follow up**
  - **Serial USG / Growth, amnios**
  - **Fetal well being**

- **Delivery**
  - **Pediatric Surg**
  - **37 gw ↓ / No benefit**
  - **C/S?**
    - **Giant Omph- C/S**
    - **Small – Vaginal?**

Gastroschisis

- 1 / 2000- 5000 pregnancy
- Full-thickness cleft, usually to the right of the umbilical insertion
- Abdominal viscera herniate into the amniotic fluid not surrounded by a membranous sac
- Bowel coated in an inflammatory fibrous peel resulting in thickening of the bowel wall

Table 2—Risk factors in the development of gastroschisis

Risk factor group	Risk factor	References
Teratogens	Maternal smoking	(Goldbaum *et al.*, 1990; Haddow *et al.*, 1993)
	Alcohol	(Torfs *et al.*, 1998)
	Cocaine	(Hume *et al.*, 1997)
	Aspirin	(Torfs *et al.*, 1996; Martinez-Frias *et al.*, 1997)
	Ibuprofen	(Torfs *et al.*, 1996; Martinez-Frias *et al.*, 1997)
	Pseudoephedrine and ephedrine	(Torfs *et al.*, 1996)
	Phenylpropanolamine	(Torfs *et al.*, 1996)
	Methylene-dioxymethamphetamine (MDMA)	(Werler *et al.*, 2003)
Epidemiological associations	Young maternal age	(Torfs *et al.*, 1998)
	Low socio-economic status	(Torfs *et al.*, 1998)
	Poor maternal educational attainment	(Torfs *et al.*, 1998)
	Change in paternity	(Chambers *et al.*, 2007)
	Short cohabitation time	(Chambers *et al.*, 2007; Rittler *et al.*, 2007)
	Poor maternal diet	(Torfs *et al.*, 1998)

Mastroiacovo P, Lisi A, Castilla EE, *et al.* 2007. Gastroschisis and associated defects: an international study. *Am J Med Genet* **143A**: 660–671.

- Associated Anomalies
  - **12% / ICBDR 3322 GS**
  - **5-50%**
- Chromosomal Abnormality
  - **1.2% / ICBDR**
  - **No karyotype**
- Syndrome
  - **0.7% / ICBDR**

- Bowel Complications
  - **Intestinal atresia**
  - **Stenosis**
  - **Perforation**
  - **Necrosis**
  - **Volvulus**
  - **Short bowel syndrome**

  Complex Gastroschisis

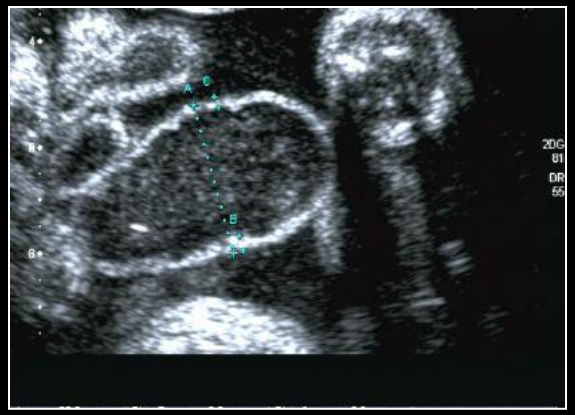
- **10 % Atresia**
  - **Omfolomesenteric vessel**
  - **Amniotic fluid ?**

Prenatal Diagnosis

- Easy
- 11-14 GW/ 70-100%
  - 9-11 gw / physiological herniation
  - <12-14 gw/ prenatal diagn?
- 18-24 GW / 90-100%

- Detailed USG
- Karyotype?

- **Bowel**
  - **Dilatation**
  - **Wall thickness**
  - **İntraabdominal bowel dilatation**
- **Stomach**
  - **Dilatation**
- **Polihidramnios**



Is complex gastroschisis predictable by prenatal ultrasound?

M Kuleva,[a] N Khen-Dunlop,[b] Y Dumez,[a] Y Ville,[a,c] LJ Salomon[a,c]

BJOG 2012;119:102–109.

Reference	*n*	Prenatal UM evaluated	Adverse outcome (*n*)	Prenatal UM predictive of outcome	OR (95% CI)	*P* value
Bond 1988[29]	11	BD (no threshold) Thickened BW Defect size Time of exposure to amniotic fluid	Neonatal GI complications (3)	Combination of BD and thickened BW	NR	NR
Langer 1993 [14]	24	BD >18 mm	Bowel resection (12)	–	7.86 (0.61–220)	NS
Babcook 1994[15]	24	BD Thickened BW	Neonatal GI complications (9)	BD >11 mm	NR	<0.05
Alsulyman 1996[18]	21	BD ≥17 mm	Bowel resection (2)	–	NR	NS
Abuhamad 1997[35]	17	SMA Doppler BD >10 min	Bowel resection Staged repaired Prolonged hospital stay	BD	NR	<0.01
Japaraj 2003[17]	45	BD ≥17 mm IUGR Thickened BW AFI	Neonatal GI complications (12) ND (6) Combination (15)	Polyhydramnios	∝ (2.15–∝)** 9.25 (0.67–143)** ∝ (1.48–∝)**	0.003 0.024 0.002
Piper 2006[19]	27	BD >6 mm	Readmission for bowel obstruction (4)	BD	NR	<0.001
Nick 2006[20]	58	IBD >6 mm IUGR Oligohydramnios UA Doppler	Small bowel atresia (10)	IBD IUGR	∝ (54.2–∝)** 5.7 (1.12–30.9)**	<0.0001 0.019
Badillo 2008[33]	64	Present of any additional GI abnormality (IBD,EBD, thickened BW, stomach herniation, echogenic bowel, loss of peristalsis)	CGS (11) ND (3)	None	–	NS

Reference	*n*	Prenatal UM evaluated	Adverse outcome (*n*)	Prenatal UM predictive of outcome	OR (95% CI)	*P* value
Davis 2009[36]	25	BD >17 mm Thickened BW	CGS (NR)	None	–	NS
Nicholas 2009[37]	80	BD ≥10 AFI abnormalities IUGR Stomach dilatation Hyperperistalsis	Composite: ND, prolonged DHS, >2 surgeries, sepsis, bowel atresia (NR)	IUGR Hyperperistalsis	5 (0.9–29.1) 2.7 (1.0–7.3)	0.05 0.07
Payne 2009[38]	89	BD	Neonatal GI complications (27)	BD >1 0mm	NR	0.01
Garcia 2010[39]	94	EBD	Neonatal GI complications (8) IUFD (5) ND (5)	EBD ≥25 mm	4.08 (1.04–16.1) None None	0.003 NS NS
Contro 2010[21]	48	IBD >6 mm EBD >6 mm	Bowel obstruction (8)	IBD	4.05 (1.12–14.7)	0.037
Huh 2010[22]	43	IBD (no threshold) EBD (no threshold)	Neonatal GI complications (8)	IBD of multiple bowel loops	∝ (7.52–∝)**	0.001
Alfaraj 2011[34]	98	Gastric dilatation	Neonatal GI complications (7)	–	–	NS
Current study	105	SGA IBD >6 EBD >6 Stomach dilatation Stomach herniation Thickened BW	CGS (14) IUFD (2) ND (2)	IBD None None	4.13 (1.32–12.90)	0.018 NS NS

- **Prognosis**
  - **Bowel comp. / 30%**
  - **Survive ~ 90%**

Complication type	Incidence/range	Authors
Uncomplicated	69–90%	Molik *et al.*, 2001; Eggink *et al.*, 2006; Arnold *et al.*, 2007; Jager and Heij, 2007; Vachharajani *et al.*, 2007
Complicated (including bowel atresia, volvulus, etc)	10–31%	Molik *et al.*, 2001; Eggink *et al.*, 2006; Arnold *et al.*, 2007; Jager and Heij, 2007; Vachharajani *et al.*, 2007
Time to full enteral feeds in primary closure	26–30 days	Swartz *et al.*, 1986; Driver *et al.*, 2000; Curry *et al.*, 2004
Risk of early necrotising enterocolitis	4–10%	Snyder, 1999; Eggink *et al.*, 2006
Functional bowel dysfunction in later life	50%	Tunell *et al.*, 1995
Risk of short-bowel syndrome	Up to 5%	Driver *et al.*, 2000; Molik *et al.*, 2001

David et al, Prenat Diagn, 2008

- **Abortus- IUD 5-10%**
- **Preterm delivery 5-8%**
- **FGR 30-50%**

- **Follow-up**
  - **Amnio-exchage / Not usefull (Midrio et al,2007)**
  - **Seri al USG / Fetal growth, Amnios, Bowel dilatation**
  - **Fetal well being / CTG, UA Doppler**

- **Delivery**
  - **Pediatric surgeon**
  - **37 gw ↓/ ? No benefit**
  - **C/S?**
    - **Mostly C/S**

Gastroschisis – what should be told to parents?

Karine Lepigeon[1], Tim Van Mieghem[2], Sabine Vasseur Maurer[3], Eric Giannoni[4] and David Baud[1]*

Prenatal Diagnosis 2014, 34, 1–11

Table 1 Neonatal outcomes related to mode of delivery

References	Study period	Duration (years)	n patients included	VB/CS	Bowel outcomes (all NS, except*)	Primary closure (all NS, except*)	Sepsis (all NS)	Length of hospital stay (days) (all NS)	Mortality (all NS)
Baud et al. (2013)[38]	1980–2011	31	261	191/70	Bowel damage 16% vs 16%	82% vs 73%	27% vs 34%	46.7 vs 50.3	4% vs 7%
Snyder et al. (2011)[77]	1999–2009	10	149	69/80[a]	NEC 4.4% vs 5.0%			39 vs 44	4.3% vs 5%
Boutros et al. (2009)[78]	2005–2007	2	192	119/73		92% vs 88%	18% vs 28%	47.3 vs 57.1	2.6% vs 8%
Abdel-Latif et al. (2008)[79]	1997–2005	8	631	343/288[b]	NEC 2.3% vs 1.4%		20% vs 18%		8.7% vs 5.9%
Hadidi et al. (2008)[80]	1986–2006	20	86	23/23[c]	Bowel complications 70% vs 35%*	65% vs 87%*	36% vs 26%	61.6 vs 51.6	4.3% vs 8.7%
Snyders et al. (2005)[81]	1970–2003	33	222	119/103	NEC 5% vs 3,9%	73% vs 59%	26% vs 17%	40.3 vs 39.5	8.4% vs 5.8%
					Atresia 10% vs 10.7%				
					Bowel complications 32.8% vs 18.4%*				
					Bowel obstruction 11% vs 8.1%				

Table 1 Neonatal outcomes related to mode of delivery

References	Study period	Duration (years)	*n* patients included	VB/CS	Bowel outcomes (all NS, except*)	Primary closure (all NS, except*)	Sepsis (all NS)	Length of hospital stay (days) (all NS)	Mortality (all NS)
Salihu et al. (2004)[83]	1983–1999	16	354	174/180					6.9% vs 3.3%
Singh et al. (2003)[84]	1990–2000	10	181	102/79	Bowel obstruction 8% vs 11%		13% vs 14%	25 vs 30	4.9% vs 3.8%
Strauss et al. (2003)[85]	1989–1999	10	60	29/31	Bowel damage 32% vs 31%				6% vs 7%
Vilela et al. (2001)[86]	1995–1999	4	31	17/14					58.8 vs 42.9%
Driver et al. (2000)[87]	1991–1997	6	91	67/24				44.7 vs 33.6	
Haw et al. (2000)[88]	1988–1999	10	71	32/38	Bowel damage 0% vs 3%	62% vs 62%	12% vs 3%	40 vs 36	6% vs 3%

Karın Ön Duvarı Defekti Prenatal Tanısı Olan 24 Gebeliğin İrdelenmesi

Evaluation of 24 Cases of Prenatally Diagnosed Abdominal Wall Defects

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Turkiye Klinikleri J Gynecol Obst 2008, 18:34-40

TABLO 1: Olguların klinik özellikleri.

	Gastroşizis N= 11	Omfalosel N= 13
Yaş (ort ± SD) (yıl)	22.5 ± 5.1	26.8 ± 2.0
Nulliparite (n/N,%)	8/11, 72.7	7/13, 53.8
Tanı hafta (ort ± SD)	21.7 ± 7.5	22.0 ± 6.4
İlave yapısal anomali (n/N,%)	0	4/13, 30.8
Kromozom anomalisi (n/N,%)	0	1/13, 7.7
Canlı doğum (n/N, %)	9/11,81.8	8/13, 61.5
Doğum hafta (ort ± SD)	37.4 ± 2.1	37.2 ± 2.5
34 hafta öncesi doğum (n/N,%)	1/9, 11.1	3/8, 37.5
Doğum kilo (ort ± SD)	2243 ± 575	2217 ± 830
Intrauterin gelişme geriliği (n/N,%)	6/9, 66.6	3/8, 37.5
Sezaryen (n/N,%)	8/9, 88.8	6/8, 75

TABLO 2: Olguların perinatal sonuçları.

		Omfalosel		
		İlave kromozomveya yapısal anomali		
	Gastroşizis	Yok	Var	Toplam
	N= 11	N= 9	N= 4	N= 13
Tıbbi tahliye uygulanan (n/N,%)	1/11, 9	2/9, 22.2	2/4, 50	4/13, 30.8
Tıbbi tahliye dışındaki olgular (N)	10	7	2	9
In utero eksitus (n/N,%)	1/10, 10	0	1/2, 50	1/9, 11.1
Neonatal eksitus (n/N,%)	1/10, 10	2/7, 28.6	1/2, 50	3/9, 33.3
Sağkalım (n/N,%)	8/10, 80	5/7, 71.4	0	5/9, 55.5
Canlı doğup yaşayan (n/N,%)	8/9, 88.8	5/7, 71.4	0	5/9, 55.5
Perinatal mortalite (n/N,%)	2/10, 20	2/7, 28.6	2/2, 100	4/9, 44.4

TABLO 3: İzole karın ön duvarı defekti olan ve canlı doğan olguların neonatal sonuçları.

	Gastroşizis		Omfalosel	
	Barsak lumen >17 mm ve/veya batın içi barsak dilatasyon		Omfalosel kese büyüklüğü	
	Var	Yok	8 cm küçük	8 cm büyük
Canlı doğan (N)	6	3	4	3
Barsak komplikasyonu	1	0	0	0
Sepsis	0	0	1	1
Neonatal eksitus	1	0	1	1

Body Stalk Anomaly (Limb -body Wall Complex)

- 1/14 000- 42 000 pregnancy
  - Abdominal wall defect
  - Limb deformities
  - Kyphoscoliosis
  - Absent or short umbilical cord
  - Craniofacial defects

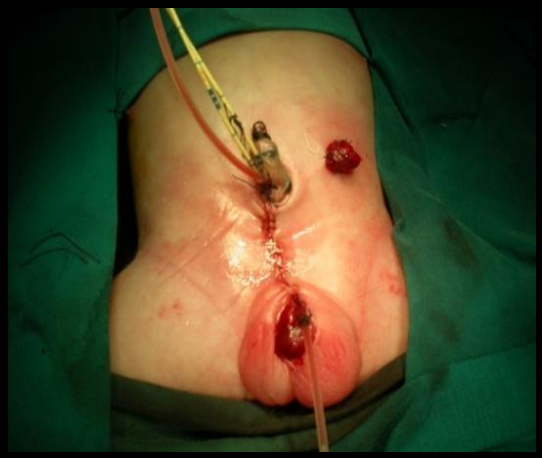
Cloacal Exstrophy

- 1/200-400 000 pregnancy
- Lower abdominal wall defect
- Bladder nonvisuable
- Echogenic bowel between bladder

Nadir Görülen Bir Karın Ön Duvarı Defekti: Kloakal Ekstrofi

A RARE ANTERIOR ABDOMINAL DEFECT: CLOACAL EXSTROPHY

Turkiye Klinikleri J Gynecol Obst 2005, 15:49-51



Ultrasound findings in fetal abdominal wall defects.

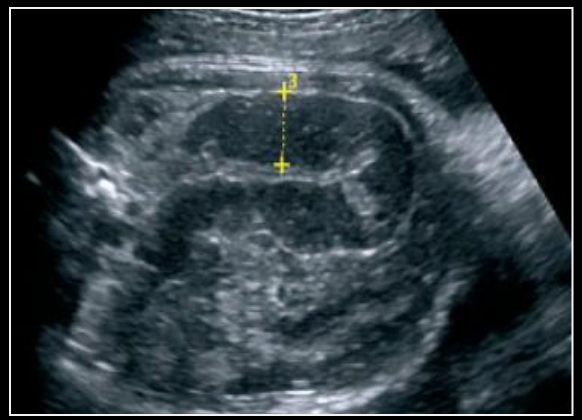
Abnormality	Covering membrane	Site of defect	Umbilical cord insertion	Additional findings
Omphalocele	Yes	Umbilical insertion	Omphalocele membrane	
Gastroschisis	No	Right of umbilical insertion	Normal insertion	
Umbilical hernia	Yes	No umbilical ring defect	Normal insertion	
Pentalogy of Cantrell	Yes	Above umbilical insertion	Omphalocele membrane	Anterior diaphragmatic hernia, sternal clefting, ectopia cordis, and intracardiac defect.
OEIS complex	Yes	Umbilical insertion	Omphalocele membrane	Bladder exstrophy, imperforate anus, and spina bifida.
Body-stalk anomaly	Herniated organs in extraembryonic coelom	Whole anterior abdominal wall	Cord absent or shortened	Kyphoscoliosis, cranial defects, and limb defects.
Bladder exstrophy	Not applicable	Below umbilical insertion	Low insertion	Non-visualisation of bladder, lower abdominal bulge (exstrophied bladder), small penis with anteriorly displaced scrotum (if male), and widening of the iliac crests.
Cloacal exstrophy	Not applicable	Below umbilical insertion	Low insertion	Renal anomalies, neural tube defect, omphalocele, vertebral anomalies, non-visualisation of the bladder, distended bladder, hydrocolpos, dilated or echogenic bowel, umbilical cord cyst, separated pubic bones, and 'elephant trunk' sign.

Bowel

- **Small bowel**
  - **Lumen<7mm**
  - **Peristaltism+**
- **Large bowel**
  - **Lumen<18mm**
  - **Peristaltism-**

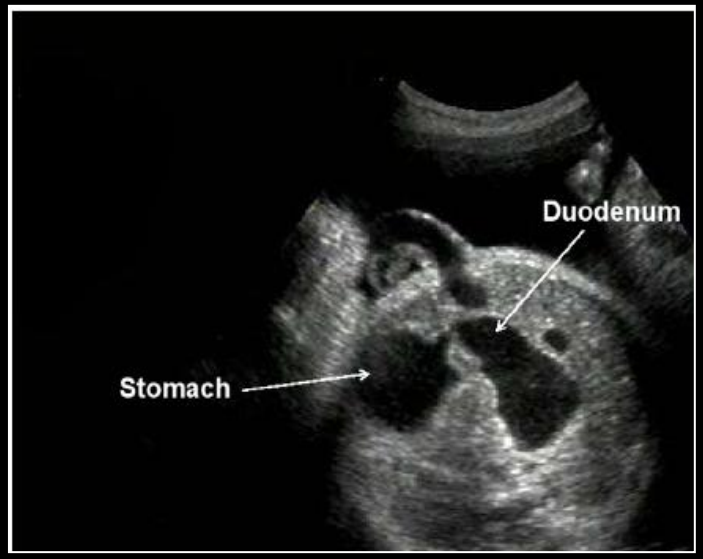
Table 4. Small bowel lumen size at different gestational age according to Parulekar[28]

Gestational age (weeks)	Average small bowel lumen size (mm)
>40	4.4
35–40	3.7
30–35	2.9
25–30	1.8
20–25	1.4
15–20	1.2
10–15	1.0



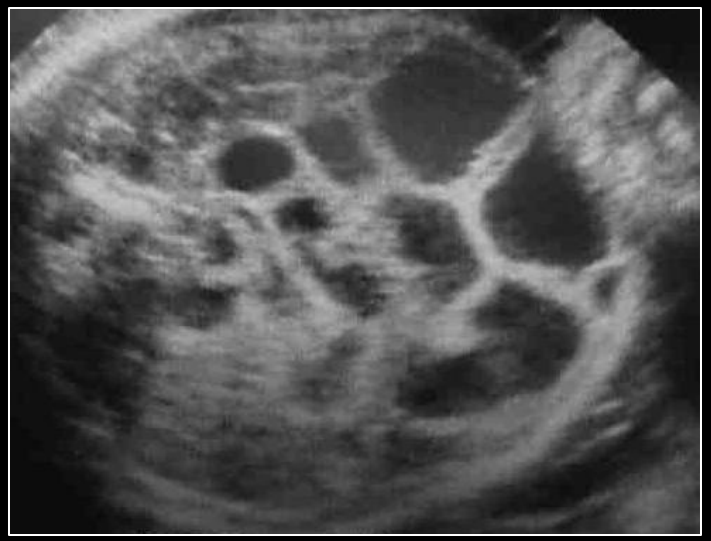
Duodenal Atresia

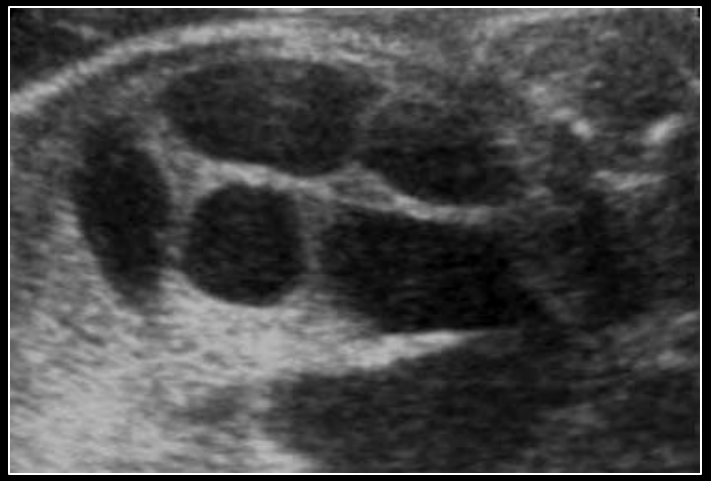
- 1/5000 pregnancy
- “Double-bubble”
- Polihydramnios
- Tr 21 / GI anomalies ,20-40 %
- Isolated / Survive 95 %



Jejunoileal Atresia

- 1/3000 pregnancy
- Dilated bowel
- Polihydramnios
- Cystic fibrosis test
- Survive ˜ 90%





Meconium ileus

- Dilated bowel
- Calcification, hyperechogenic
- Ascites
- Cystic fibrosis

Volvulus

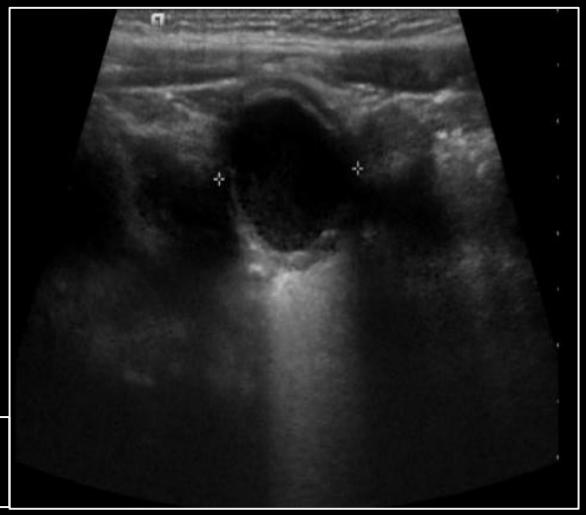
- “kinked” loop
- “whirlpool”
- Acute condition

Intraabdominal Cyst

- Omental

- Mesenteric

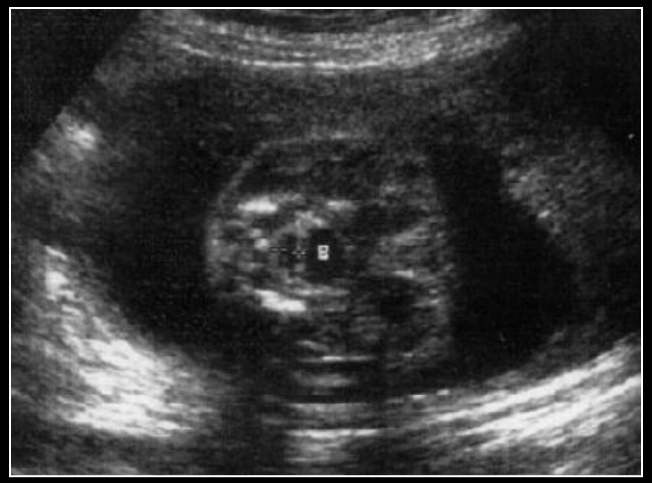
- Ovarian



- Duplication

Anorectal Anomaly

- 1/5000 pregnancy
- Dilated bowel
- Late, very difficult
- Associated anomalies (VACTERL, Cloaka …)



Fetal MR

Ana Karnında
Manyetik Rezonans Görüntüleme

Turkiye Klinikleri J Radiol-Special Topics 2011;4(2):52-60

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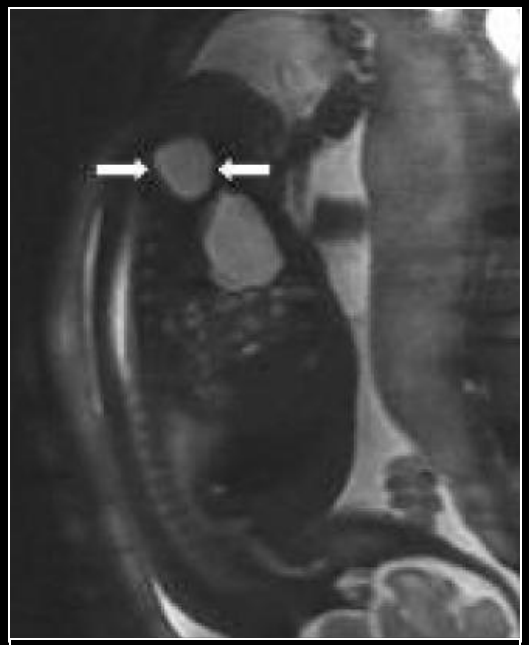
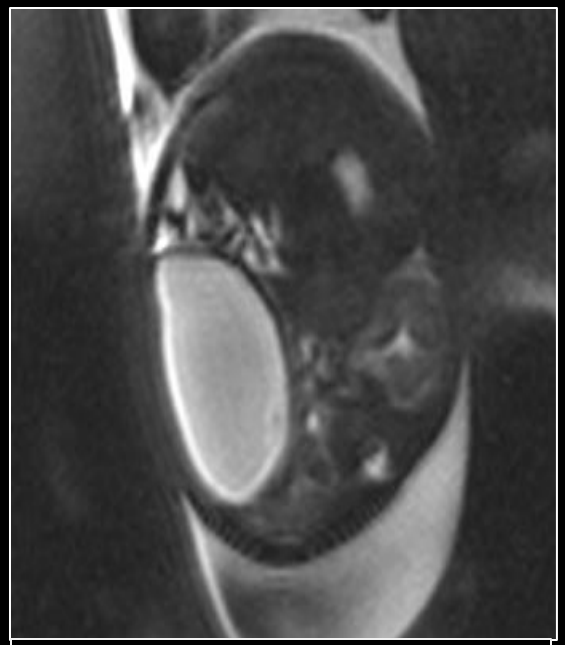
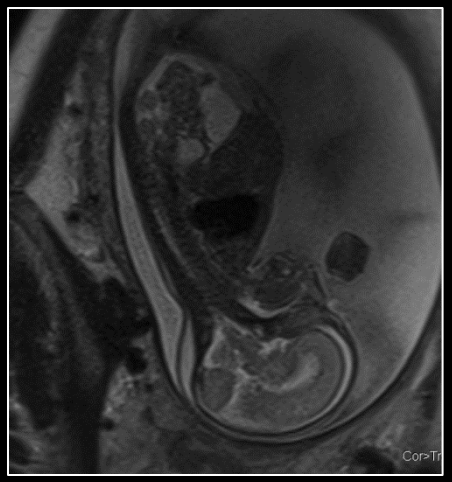


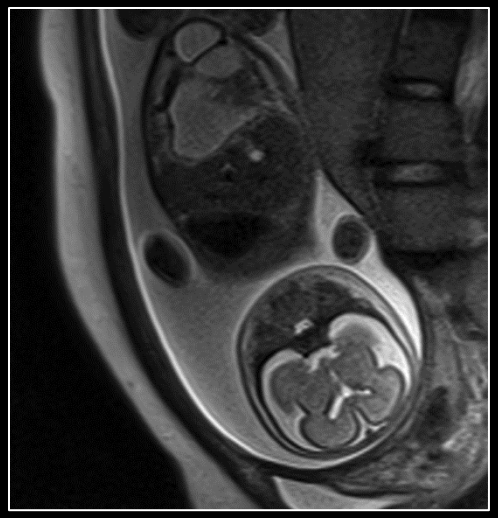
Rektal Dublication



Intestinal Dublication

Ovarian Cyst



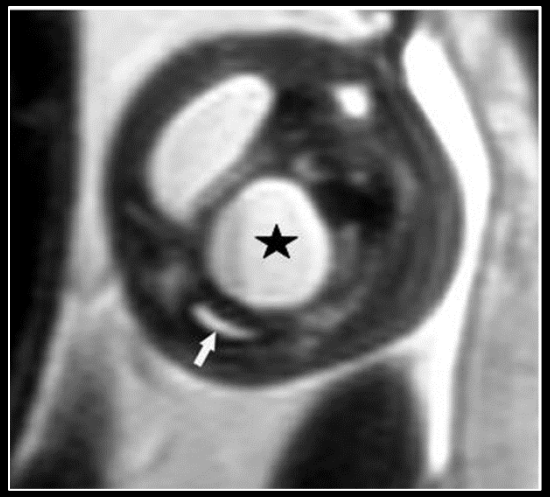


Duedonal Atresia

Cloacal Anomaly

Congenital Imperforate Hymen with Hydrocolpos Diagnosed Using Prenatal MRI

Adaletli et al. *AJR* 2007; 189:W23–W25



Teşekkürler