B D R News

The official newsletter of the Birth Defects Registry of India,
(A unit of Fetal Care Research Foundation)

Volume 5 Issue 1:Jan 2005

PROCEEDINGS OF THE BDR MEETING HELD ON 29/01/05

The first BDR meeting of the year was held on 29/01/05. The CME on "Fetal Gastro Intestinal Tract Anomalies" kicked off with a peppy note from the director Dr. S. Suresh. His gut feelings made him assure the audience an interesting day long scientific session. He also called it a dynamic knowledge acquisition program which throws light on the concerned subject and helps the doctors deepen their knowledge and confidence regarding the management of birth defects. The CME was designed and supervised by Dr. Sujatha Jagadeesh, Consultant Dysmorphologist, BDRI. The excerpts of the presentations given by Dr. Suseela Vavilala, Dr. Radhika Ramesh and Dr. Bindu Sridevi—Fellow Trainees, Fetal Medicine, Mediscan Systems and Dr. S. Balagopal, Consultant Paediatric surgeon, Sri Ramachandra Medical College & RI, Chennai, are given below.

SONO EMBRYOLOGY OF GASTROINTESTINAL TRACT (GIT) - Dr.Radhika Ramesh

Dr. Radhika in her talk, dealt in detail about the various stages of development of the gut. She said that, majority of development of the gastrointestinal tract occurs between 4-8 weeks after fertilization. By the fourth week, folding of the cephalic, caudal and lateral ends of the embryo occurs giving the embryo a human form. During this process of folding, the yolk sac gets incorporated as primitive gut from which the gastrointestinal tract develops. Primitive gut is divided into 3 parts, namely, the foregut, midgut and the hindgut. **Foregut** lies between the brain and the heart. The oropharyngeal membrane separates the heart from the stomatodeum. Foregut gives rise to 1)pharynx and its derivatives, 2) upper and lower respiratory system, 3) oesophagus and stomach up to proximal duodenum, 4) liver and biliary apparatus. **Esophagus** develops from foregut immediately caudal to pharynx. The partitioning of trachea from the esophagus is by tracheo esophageal septum. The epithelium proliferates and fills the lumen and recanalisation occurs later. If separation of esophagus from the laryngotracheal tube is incomplete, Tracheo Esophageal Fistula (TEF) results.

Incomplete canalization leads to Esophageal Atresia. **Stomach** develops by the dilatation of the distal part of the foregut. As it assumes adult shape it rotates 90 degrees clockwise around the longitudinal axis and assumes its final position. Anomalies of the stomach are uncommon. **Duodenum** develops from the caudal part of the foregut and cranial part of the midgut. The two parts are joined distal to the origin of bile duct. Duodenum along with the stomach rotates and lies retroperitonially. Duodenum and the rest of the small bowel are filled with proliferating epithelium. Recanalisation later occurs for about 3 weeks. Failure of recanalization results in Duodenal Atresia. Most atresias involve the second and third part of the duodenum, distal to bile duct.The two parts are joined distal to the origin of bile duct. Caudal part of the foregut develops into liver, pancreas and spleen. **Mid gut** forms from the lateral folds. As the abdominal walls form, part of endoderm germ layer is incorporated into the embryo as midgut. This gives rise to 1)rest of duodenum,2) small intestine, and 3) large bowel up to two thirds of transverse colon**.The rotation of the midgut is the most crucial event.** The midgut projects into the umbilical cord by 6 weeks, rotates along the axis of artery and then returns to the abdomen by 10 weeks, again rotates, and elongates to form the loops of small bowel which lies in the centre. The large bowel and caecum occupy the right side of the abdomen Errors in midgut rotation can give rise to various anomalies like midgut volvulus. **Hindgut** is formed by the folding of the caudal end of the neural tube. The terminal part of the hindgut dilates to form cloaca. Hind gut gives rise to left one third of colon, descending colon, sigmoid colon, rectum and superior part of anal canal. Partitioning of the cloaca occurs by urorectal septum, which separates the rectum and anal canal dorsally and urogenital sinus ventrally. The inferior one third of the anal canal forms from the proctodeum. Errors in caudal folding result in Cloacal Exstrophy / Bladder exstrophy. Other hindgut anomalies include Hirschsprung disease, Anal agenesis, Anal stenosis due to failure of the anal membrane to perforate during the 8th week and Rectal atresia due to abnormal recanalisation of the colon. Errors in embryonic folding:1)Failure of lateral folds to meet in midline may result in omphalocele; 2) Failure of ventromedial migration of paired mesodermal folds can result in Pentology of Cantrell and 3)Vascular compromise of umbilical vein/omphalomesentric artery results in Gatroschisis. After explaining the development of the gut, Dr. Radhika briefed the audience about routine ultrasound imaging of the fetal gut

IMAGING OF THE FETAL GIT

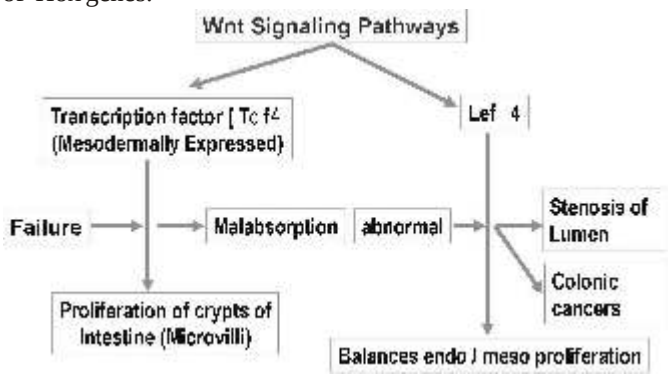
Esophagus: Esophagus need not be imaged routinely, but it can be imaged when looked for, especially after swallowing. It can be imaged in the anterior coronal plane, anterior to the descending thoracic aorta. But the **stomach** can be imaged with ease and can be seen as early as 9 weeks. It can be viewed easily from 14 weeks in the left side of the fetal abdomen. If stomach is not imaged at the first instance, the patient can be rescanned after few hours. Liver occupies most of the fetal abdomen, but the margins are indistinct, similar to spleenic borders in the early weeks. As the gestational age advances, the margins become sharp. Gall bladder can be imaged in the right upper quadrant, as drop shaped structure Small bowel is imaged centrally in then shaped structure along the inferior border of the liver Small bowel is imaged centrally in the mid and lower abdomen. Bowels loops become more distinct as the fetus grows. Active peristalsis is observed in the

small bowel from 14 weeks in the left side of the fetal abdomen. If stomach is not imaged at the first instance, the patient can be rescanned after few hours. **Liver** occupies most of the fetal abdomen, but the margins are indistinct, similar to spleenic borders in the early weeks. As the gestational age advances, the margins become sharp. **Gall bladder** can be imaged in the right upper quadrant, as drop shaped structure along the inferior border of the liver **Small bowel** is imaged centrally in the mid and lower abdomen. Bowels loops become more distinct as the fetus grows. Active peristalsis is observed in the small bowel. **Large bowel** is seen in the periphery of the abdomen as tubular structure. It is hypoechoic as compared with small bowel due to the presence of meconium. Large bowel peristalsis is not observed in the fetus. **Rectum** can also be imaged.

GENETICS OF GIT - Dr.V. Bindu Sridevi

Dr. Bindu laid a good foundation for the day's program by enlightening the audience about the recent advancements in the genetics of GIT. She emphasized the fact that professionals need to equip themselves with the knowledge on genetics to give proper information to patients, once they make a diagnosis of an anomaly. Such knowledge facilitates better management of the problem, during pregnancy and after delivery.

Embyology: Newly discovered genes called the Homeobox genes play an important role in the formation of GIT. It was first found in DrosoPhila Fly and it was originally thought to be important for the development of the central nervous system. Homeobox genes(HG) occur in 4 clusters called the Hox genes and are found on chromosomes 2.7,12 & 17. The concentration of the Hox proteins in the chromosomes and the location of production in the developing embryo and the timing with which they become active in the body plan are the key determining factors in the development of the gut. The target gene specificity crucially affect the former three, for if a given Hox gene sequence is altered by mutation, the resulting protein that is expressed may not bind to its target genes. For example, the diagram below explains one of the pathways of the mechanism of Hox genes.



Transcription factor Tc 4 responsible for the proliferation of microvilli, when interrupted by terotogens like beta carotene, can result in malabsorption disorders like stenosis of the gut lumen, colonic cancer etc. Another factor L 4 balances the proliferation of endodermal and mesodermal cells also produces the same effect when there is an insult.Other type of signaling is called the Hedgehog signal where a balance between two factors called Shh and BmP4 also determine the proliferation of the cells. Once the cells develop, vasculogenesis of the cells occur by the aggregation of angioblasts which helps in the vascular supply of the gut. If this process is affected by terotogens, it may result in Gastroschisis. The neural crest cells then migrate and innervate the gut. Any disruption here can cause disorders like Hirschsprung. After all these, the gut organs arrange themselves on the respective side of the body. All symmetric organs like limbs are placed laterally and asymmetric organs are placed in the middle. Positioning is also controlled by a set of genes. Mutations of these genes lead to Heterotaxy Syndromes.

Dr. Bindu also talked briefly about the various genetic disorders concerning the gut such as TEF,duodenal atresia, jejunal, ileal atresias, large bowel atresia, omphalocele, gastroschisis ,cystic fibrosis and diaphragmatic hernia.. Once a genetic diagnosis is made the etiology i.e whether it is a chromosomal, genetic or sporadic problem should be looked for to facilitate proper genetic counseling.

Discussion

• Does genetic testing play a role in Cystic Fibrosis?

If a woman had an index child with the history of Cystic Fibrosis, further investigation can be done. A bright bowel otherwise should not raise an alarm of only Cystic Fibrosis as it has multivaried etiology and this need not always present as echogenic bowel.

• If the index child's sweat chloride is positive, how do you go about prenatal diagnosis of Cystic Fibrosis?

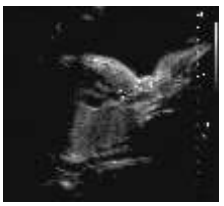
When sweat test is positive, carrier testing is done for the parents to find out the mutation for Cystic Fibrosis and once it is confirmed, the fetus is tested by invasive test to see whether it is affected.

• How do you differentiate between single gene and chromosomal disorder?

Single gene disorder repeat in generations unlike chromosomal disorders. The karyotype in single gene disorders would be normal. Drawing up a three generation pedigree of the family helps to find out the transmission of the problem to conclude on recessive or dominant mode of inheritance.

BOWEL MYSTERIES & CASE SCENARIOS - Dr. Susila Vavilala

Dr. Susila Vavilala presented the mysteries of various disorders of the gut, through interesting case scenarios. The cases are as follow:



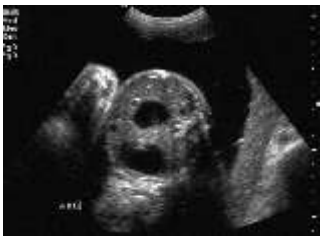
Case I: 26 years old mother with an obstetric history of G3P2L1 at 34 weeks of gestation was referred for polyhydramnios. Her second pregnancy was terminated due to Arnold Chiari defect in the fetus. Her ultrasonography (USG) findings were a small tubular, unechoic area with amniotic fluid index- 40 and biometry corresponding to gestational age. Diagnosis was esophageal atresia.

Stomach is visualized well after 12 weeks. Absent or small stomach reflects a relative obstruction to the passage of amniotic fluid. The causes for ineffective swallowing and small stomach could be esophageal atresia, fetal goiter pressing the esophagus, facial

clefts, CNS anomaly, oligohydramnios and congenital microgastria. One should also remember that 1% of normal fetus can have a small stomach. When small stomach is seen, rescanning is mandatory. It is also important to measure the liquor volume and look for associated anomalies like VATER/VACTERL. ***A tubular unechoic area and persistent non visualization of the stomach are suggestive of esophageal atresia.***

Case II:36 years old mother with G2P1L1 obstetric history and II degree consanguinity was referred for ? Ileal atresia. Her USG findings were two unechoic areas with double bubble sign, large stomach, Tetrology of Fallot defect, low placed multicystic dysplastic right kidney, single umbilical artery and polyhydramnios. The diagnosis was **Down syndrome** which was confirmed by fetal karyotyping and the fetus had IUFD. Fetal autopsy confirmed the USG findings. ***Large stomach without other findings may be suggestive of gastric outlet obstruction or volvulus, pyloric stenosis, duodenal atresia or annular pancreas.***

Case III: 24 years old primi mother was referred for polyhydramnios at 34-35 weeks. Her USG findings were double bubble sign suggesting duodenal atresia and other systems were normal. She delivered a male baby with 1.8 Kgs and good APGAR. Postnatal cord blood tested proved normal karyotype.



The child underwent duodeno duodenostomy after 24 hours. ***Intra operative diagnosis was annular pancreas.*** While antenatally diagnosing this problem, fetal stomach should be scanned only in transverse plane as fetal stomach in oblique plane may mimic double bubble sign. ***Two cystic spaces maintaining continuity with the stomach and polyhydramnios is diagnostic of duodenal obstruction.*** Theoratically duodenum can be seen at 11weeks. But it can be visualized only after 24 weeks in practice. Duodenal atresia with or without esophageal atresia warrants fetal karyotype and fetal echo as there is 50% association with Down syndrome.

Case IV: 24 years old primi mother with III degree consanguinity was referred at 23 - 24 weeks in view of polyhydramnios. Her USG finding was dilated small bowel. On repeat scans at 2 weekly intervals , it disappeared. ***In practice when a dilated small bowel is seen, the diameter of the bowel should be measured. The normal diameter of the small bowel is 5mm and the large bowel is 23 mm. If it is > 5mm, repeat scan is mandatory. One should also look for peristalsis, liquor volume and the length of the segment involved.***

Case V: 20 years old primi mother on routine scan at 28 weeks of gestation showed hyperechoic bowel. Normally fetal bowel is imaged between two unechoic spaces i.e between the stomach and the bladder. Small bowel is more echogenic than the liver and less than that of the iliac bone. Hyperechoic bowel may occur due to vascular compromise causing decreased bowel function leading to hyperperistalsis, reduced water content and inspissated meconium. This could be a nonspecificfinding in a normal fetus. But this is also associated with aneuploidy, IUGR & IUFD(11%), bowel atresias / obstructions, cystic fibrosis and congenital infections. Hyperechoic colon due to meconium is normal in the third trimester, whereas it is abnormal if seen in small bowel. One should be cautious to assess the echogenicity, as by increasing the echo setting of the machine even a normal bowel can appear echogenic. Hyperechoic bowel increases the risk for Down syndrome by 3%. Hence high risk pregnancies with echogenic bowel with advanced maternal age/ abnormal maternal screening tests/associated anomalies warrant karyotyping, serological studies for infection and DNA testing for cystic fibrosis. Low risk mothers with isolated finding need to be followed up.

Case VI: 20 years old primi mother with III degree consanguinity was referred at 30—31 weeks for a cystic leision in the abdomen. Her USG revealed multiple unechoic spaces with dilatation of bowel loops, non visualization of large bowel, liquor in the upper limit of normal volume all of which were suggestive intestinal obstruction involving the small bowel. Generally mid abdominal loops are seen here with hyperperistalsis. Ascites and polyhydarminos are the associated findings. ***This situation warrants serial ultrasound imaging. The causes could be Jejunal/Ileal atresia, intestinal volvulus, meconium ileus and other functional obstruction.***

Case VII: Low risk primi at 36 weeks of gestation was referred for reduced fetal movements for a scan. She was asked to get a bio physical profile done. Her USG showed multiple dilated loops suggesting intestinal obstruction. She had a caesarean delivery and the baby was operated immediately after 2 days. Bowel gangrene was detected and excised. The diagnosis was intestinal volvulus. This appears as dilated loops antenatally with absent peristalsis, intraluminal sludge and ascites. Volvulus is caused due to failure of the intestines to return to the abdomen or due to improper fixation of the mesentery. This occurs at or near the duodeno jejunal junction and can be detected as early as 16 weeks of gestation. ***In case of dilated bowel loops one should look for the length and diameter of the segment involved, peristalsis and liquor volume. Rescanning is necessary to correctly identify the problem.***

Case VIII: 20 years old mother with an obstetric history of G2P1L1 was referred at 29-30 weeks for a second opinion for ? Meningomyelocele. Her USG findings were, spinabifida with sacral meningocele, bilateral agenesis, prominent distal colon suggestive of ***anorectal atresia.*** This may present as dilated large bowel, blind ending rectal pouch, calcified intraluminal meconium and normal or reduced liquor. The various types of abnormal colon are, enlarged colon, small colon, anorectal atresia, ileal atresia, Hirschsprung disease, meconium ileus, meconium plug syndrome and Extrinsic compression. Dr. Susila concluded saying that ultrasonography plays an important role in the identification of fetal bowel pathology unlike in adults for the simple reason it is gasless. They can be identified as a dilated bowel segment, usually proximal to the obstruction. Dilatation may be due to bowel atresia involving any region of the gut and intestinal obstruction, volvulus and malrotation. The other condition which needs to be considered is hyperechoic bowel, where the echogenicity is comparable to that of the iliac bone. The etiology is variable and the common cause is chromosomal aneuploidy. Echogenic bowel increases the risk of aneuploidy by 3%. Though USG can provide useful information on bowel pathology, conclusive evidence can not be drawn to assign a diagnosis. Hence, postnatal studies of mysterious looking bowel are essential for accurate diagnosis and further management.

Discussion:

• If a dilated bowel is seen on scan, can it be assigned a differential diagnosis or just call it a dilated bowel?

Depending on the involvement, it may be called single dilated bowel or if many segments are involved it may be called intestinal obstruction.

•Do all babies with malrotation get into problem?

Not all babies with malrotation get into problem. It may remain as a benign condition. It is seen in different age groups in routine paediatric practice. Intervention is needed only when it presents with symptoms. No preventive surgery is required.

• Will the fetus suffer from volvulus ? If so how will it express?

When the bowel becomes obstructed or gangrenous, fetus may experience pain and movements may reduce when situation gets moribund.

FETAL ABDOMINAL CYSTS: - Dr. Bindu Sridevi

Dr Bindu while talking about abdominal cysts said that we do come across diagnosing cysts in the abdomen of the fetus, As the types and number of cysts are wide, one should have clear cut knowledge about the cysts to narrow down the differential diagnosis. Though urinary cysts are also seen along, they are not to be dealt here. Various cysts in the gut organs comprise of :

- Simple cyst, lymphangiomas, choledochal cyst and varix of umbilical vein - Liver
- Duplication cyst, mesenteric cysts, meconium pseudocyst - bowel
- Follicular cycts - Ovary
- Hydrometrocolpos - Uterus / Vagina

When a cysts is seen in the fetal abdomen, many factors such as the gestational age at presentation, location of the cyst, gender, terotogenic influence, morphology, whether it is vascular, and the associated factors need to be considered to clinch on the diagnosis.It is mandatory to postnatally evaluate the prenatal findings for further management.

Choledocal cyst: Choledocal cyst is an abnormal dilatation of the bile duct, which may be due to abnormal insertion of the bile duct into the pancreatic duct where all the pancreatic enzymes are reflected in to the bile duct causing dilatation of the same. The reasons attributed for this are the weakness of the wall and increased pressure in the bile duct.

Diagnosis:

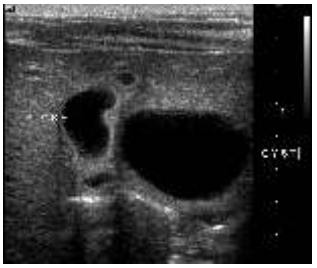
The earliest detected choledocal cysts is at 15 - 16 weeks of gestation. It is seen as a cystic mass in the right upper quadrant of the abdomen. If there is any tubular connection to hepatic/biliary ducts , it could be called a choledocal cysts conclusively. If these cystic structures do not enlarge on serial scans, postnatal evaluation is warranted to rule out congenital biliary atresia. Complications that could arise on a long term in this case would be, cholestasis, cholangitis, biliary cirrhosis , pancreatitis causing portal hypertension and hepatic failure.

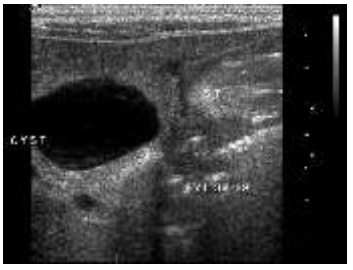
Management:

Postnatal evaluation is necessary to confirm diagnosis and if the baby presents with Jaundice and abdominal pain, surgical intervention is needed. Studies prove that long term outcome is good when surgery is instituted at the earliest possible period before complications arise. Treatment of choice is excision and construction of a Roux-en-Y hepaticoenterostomy. If excision is delayed temporary aspiration of the cysts could be done to avoid obstruction.

Enteric Cysts:

These are duplication cysts from GIT caused due to faulty recanalisation or failure of separation of notochord from ectoderm. It is necessary to look for vertebral associations like Diastemetamyelia, when a duplication cyst is found.





Diagnosis: These could diagnosed as early as 12 weeks of gestation. It can occur in both the sexes, more frequent among the males. It is commonly found in the stomach and the bowel. It may be cystic or tubular type and may be found in the esophagus, jejunum, ileum, colon and duodenum. Distinct peristalsis seen in the cyst help in diagnosis.

Management: According to studies done, thoraco abdominal duplications were the most complicated and responsible for mortality and morbidity. Surgical complications were related to the size, location, communication with GIT/vertebral canal, presence of heterotrophic gastric mucosa and involvement of mesenteric vessels. In fetuses with rapid reaccumulation of fluid and recurrence of mediastinal shift, prompt placement of thoraco amniotic shunt helped in resolution of hydrops.

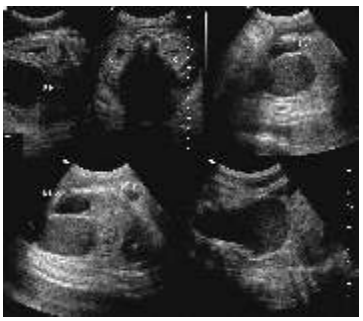
Mesenteric/Omental Cysts: Mesenteric/Omental cysts may be solitary, unilocular or multilocular with septations. They are found in both male and female fetuses. They may be seen right from the first trimester through the third trimester of pregnancy. The fluid in the cysts may be serous , bloody and mostly chylous.

Liver/Spleen Cysts: Cysts within the liver/Spleen are called the liver/spleen cysts respectively. These can be uni or multilocular in consistency.

Ovarian Cysts: Ovarian Cysts are fluid filled tumors. If a female fetus is found to have a cyst in the third trimester it may be suggestive of ovarian cyst. Earliest diagnosed is between 27—28 weeks of gestation. when the stimulation of HcG is also high on fetal ovaries. These are found more often among the Rh isoimmune mothers and those who have diabetes. Most of the tumors are benign and are uni/multilocular in consistency. If it is less than 5cm, the chances are greater for spontaneous resolution. Complications like torsion, haemorrhage, rupture or ascites and polyhydramnios can happen.

Management: Prenatally the cyst has to be monitored by serial scans. Postnatally again it should be observed for spontaneous resolution. Aspiration is indicated, if the size is >5cm in diameter and surgical incision if there is evidence of torsion or hemorrhage or if there is abdominal distension and vomiting in the child. This would preserve the fertility potential of the ovarian tissues.

Hydrometrocolpos: Hydrometrocolpos is a condition where there is dilatation of vagina/uterus due to excessive secretion or vaginal obstruction. If there is a big mass bulging out of the perineum in the lower abdomen, it is suggestive of hydrometro colpos. This occurs sporadically and is rarely associated with other genetic disorders like Mckusick Kaufman Syndrome. Uterine dilatation may result in secondary hydronephrosis with renal damage and bowel obstruction. Normal function is generally possible with surgical treatment.



Discussion: Is there a need for karyotyping when a cyst is diagnosed prenatally?

No. Even for hydrometrocolpos it is not needed. Since hydrometro colpos could be diagnosed only in the third trimester, it will not be possible to do karyotyping.

• Which type of cysts manifest earlier?

Earliest reported cysts are duplication cysts at 12 weeks of gestation. By 20 weeks omental and mesenteric cysts could be diagnosed. Later in third trimester ovarian cysts and hydrometro colpos could be diagnosed.

• Can torsion happen in utero? Is it an indication for emergency delivery?

While answering this question Dr. Suresh narrated an interesting case that was diagnosed to have an ovarian cyst. There was polyhydramnios and there was no fetal movement probably the fetus was in pain due to torsion and did not move or swallow. The cyst was aspirated and the high Estridiol of the aspirate confirmed the diagnosis. Aspiration and constant observation can help postponement of delivery.

• Does the shape of the cyst help in diagnosis?

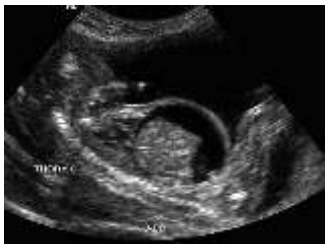
Yes, Elongated cyst suggests that the cyst is a duplicated cyst.

• Is there a true indication for intervention of abdominal mass?

If there is fetal hydrops and the mass is >5cm at III trimester or keep increasing in size and occupies the abdomen, aspiration should be done.

• How would you differentiate between a large cyst and the bladder?

The emptying and filling up of the bladder differentiates it from cysts. Also bladder fluid may be tested for Na content as opposed to cystic fluids.



FLUID IN THE ABDOMEN — Dr.Radhika Ramesh

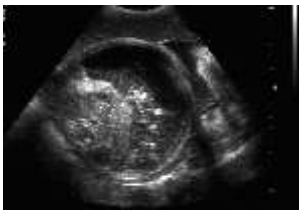
Fetal ascites: It is the fluid in the abdomen surrounding the liver, spleen and small bowel. There should be a minimum of 12-14ml in 18—20 weeks or 30 - 40ml around 30 weeks to identify it as fetal ascites. Fetal ascites is most often seen in association with hydrops. It is important to distinguish whether it is isolated or an early manifestation of hydrops. Rescanning after a week will show whether other signs of hydrops are developing.

The causes of fetal ascites could be Fetal anemia, Infections like CMV, Parvovirus,Fluid in Genitourinary tract caused by obstructive uropathy – urinary ascites, Hydrometrocolpos, Ovarian cysts, torsion/rupture,Gatrointestinal causes like perforation, Meconium peritonitis and Cardiac arrythmias,Hepatobiliary disorders and Metabolic storage disorders.

Mangement Protocol: If there is isolated ascites, structural malformations have to be ruled out. Detailed fetal echo and MCA doppler for fetal anemia and Maternal infection screen are mandatory. Fetal sampling can be done for karyotyping, fetal hemoglobin, antigen specific IgM antibodies, PCR for CMV and Parvovirus.

Therapeutic fetal paracentesis: If there is massive ascites causing thoracic compression and if the abdominal circumference is > 350mm, peritoneo-amniotic shunting may be done to facilitate vaginal delivery. If tapping is done, the fluid may be used for diagnosis. It would reveal whether the fluid is transudate or exudate. If lymphocyte count is over 90%, presumptive diagnosis of chylous ascites is made.

Calcifications in the fetal abdomen : Echogenic areas in the fetal abdomen can arise from a variety of sites and can be categorised into Peritoneal,Intraluminal,Parenchymal, Vascular and Biliary origin.It is important to distinguish from hyperechoic bowel, the acoustic shadowing should be demonstrated, and it also important to precisely determine the location of calcification.



Peritoneal calcification is almost always from meconium peritonitis. It is seen as linear opacification. Bowel perforation leads to leakage of meconium, chemical peritonitis, leading on to calcification.

Intraluminal calcifications are seen within the bowel lumen, as punctate echogenecities. Calcifications are seen proximal or distal to the site of obstruction in conditions such as anorectal atresia, volvulus, multiple bowel atresia.

Calcifications are seen either in the surface of the liver or spleen, are due to meconium peritonitis, where as intraparenchymal calcifications have a different etiology. Isolated calcifications are associated with good outcome. Multiple and widespread intraparenchymal liver calcifications warrant fetal karyotyping and workup for fetal viral infections.

Discussion:

• What should be the amount of fluid for minimal ascites?

30 ml of fluid

• Do calcification and fluid coexist?

Not necessarily.

• Does ascites need to be investigated at the first instance?

Yes. Investigation is needed to understand the underlying pathology. If it regresses on is own, testing need not be done. When there is isolated ascites, investigations should be done for fetal anemia irrespective of Rh status, infection screening and urinary ascites.

• When there is ascites and CMV is positive, Immunoflourescence & DNA studies are positive, what should be the line of management?

Fetus should be monitored for structural anomalies. Though the sequelae due to CMV infections are low, the parents have to be informed that the baby may have the sequelae. The parents through counseling are allowed to decide whether to continue the pregnancy or terminate.

ABDOMINAL WALL DEFECTS - Dr. Bindu Sridevi

Abdominal wall defects are the second most common disorder after CNS abnormality. Abdominal wall is formed by 4 ectomesodermic folds namely cephalic, caudal and two lateral folds. Abdominal wall is made up of skin, subcutaneous tissue and muscles. The most common wall defects we come across are 1) Omphalocele 2) Gatroschisis 3) Bodystalk Anomaly and 4) Cloacal/ Bladder Exstrophy. While doing a scan normal cord insertion should be made out to rule out the abdominal Defect.

Omphalocele:

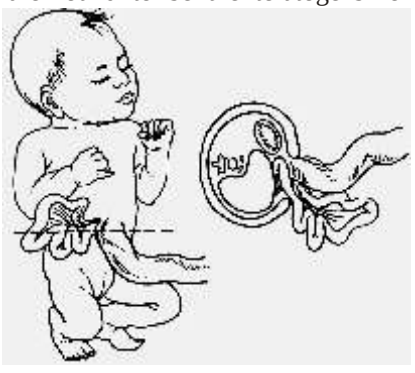
Pathogenesis & Embryology: Omphalocele is the defect in the anterior abdominal wall with extrusion of abdominal contents into the base of the umbilical cord. During embryological growth, due to lack of space the small bowel goes in to the umbilical cord and comes back to the abdomen before 12 weeks of gestation. If it stays for some reason and not gets back,

omphalocele occurs. This can be divided in to two types depending on the contents namely, intra corporeal or extra corporeal type.Small bowel is the content in the former and liver is the content in the latter type. Greater echogenicity of the small bowel and the presence of hepatic vessels help to differentiate the contents. If the diameter of the defect is > 7mm, it has to be scanned again to confirm the diagnosis. Care should be taken not to mistake physiological omphalocele before 12 weeks of age. Earliest reported omphalocele was at 10-11 weeks. This has an incidence of 2.5/10000 births and it increases with maternal age. This has also been found to be associated with a number of chromosomal anomalies like T18,T13, T21, Triploidy, Turners, Beckwith-Weidmann syndrome and Miller-Diekar syndrome.

Diagnosis: Diagnosis depends on the location of the defect which is always seen in the mid abdomen, cord insertion i.e whether attached to the apex of the defect, limiting membrane, visible ascites, contents of the sac, associated anomalies and polyhydramnios which may be secondary to intestinal obstruction and associated CNS anomalies like Holoprosencephaly. Colour Doppler is useful in differentiating haemangioma and cord hamartoma from omphalocele. Prognosis: Prognosis depends on the size, content and associated anomalies. If the defect is >5cm and even there is no associations with normal karyotype, prognosis is not good. When small bowel is the content with normal karyotype, prognosis is again bad for the fetus. Globally Gastroschisis has better prognosis than omphalocele.

Management: Studies have proved that periconceptional multi vitamin and folic acid reduces the risk of non syndromic omphalocele by 60%. However more research is needed to authenticate the effect of the vitamins. When chromosomes are normal, serial monitoring on four weekly intervals is required during pregnancy. The child has to be delivered in a tertiary set up and a Paediatric Surgeon's opinion should be sought for further management. There is no convincing evidence to support routine ceasarean section for most abdominal wall defects.

Gastroschisis: Unlike the Omphalocele, Gastroschisis is a para umbilical defect where the abdomen has a burst appearance. A disruption during vasculogenesis or error in development of the right omphalomesentric artery leading to infarction and necrosis at the base of the umbilical cord causes this problem. Smoking and cocaine are found to be the teratogens for vasucular abnormality. It is also advisable to avoid oral decongestants during pregnancy that contain Pseudoephedrine as it is a sympathomimetic vasoconstrictor. Bowel is usually the content here and bladder is very rarely seen as the content. This defect



which lacks the covering membrane is always found to the right of the umbilical cord. Gastrochisis tends to occur in younger mothers (mean age-23 years) when compared to omphalocele which has an increased incidence with maternal age(mean age-28 years).

Diagnosis: Ultrasound is the key for diagnosis during first trimester, However raised MSAFP level in the second trimester, cord insertion, thickened, floating bowel also help in the diagnosis of this defect in the ventral wall.Once it is suspected, it is advisable to do repeat scans to confirm the diagnosis. Complications associated would include bowel atresia, secondary to ischemia, perforation and fetal ascites, poly hydramnios due to obstruction and vanishing gut. If one finds gatroschisis becoming small in size during repeat scans, it may be suggestive of vanishing gut which has long term morbidity as in small bowel syndrome. Fetal distress is also a frequent complication here. When small bowel comes out, the vagal nerve gets stretched causing fetal distress and hence it is necessary to use fetal kick charts or simple home monitoring to avoid distress.Associated problems may be cardiac anomalies very rarely, amyoplasia (arthrogryposis) and bowel complications. Gatroschisis is not associated with aneuploidy.

Prognosis & Management: Prognosis depends on prematurity of the infant, neonatal sepsis and other intestinal complications. Delivery at term is appropriate for the fetus with normal appearing bowel on ultrasound. LSCS is not indicated for delivering the fetus. Vaginal delivery may be allowed. A Paediatric surgeon and a tertiary neonatal intensive care unit should be available at the delivery site. Studies show that gastroschisis minor form has a distinct survival advantage over the major form of the defect.

Body stalk anomaly: Body stalk anomaly occurs due to failure of formation of body stalk or a defect in all abdominal wall folds. This happens as a consequence of passage of the lower half of the fetus into the coelomic cavity because of partial immmobilisation as a result of short cord.

Diagnosis: If a grotesque appearing mass is seen in lower abdomen with a short umbilical cord, adherent to placental membranes and the fetus persistently facing the placenta with a sac with abdominal content, it may be suggestive of body stalk anomaly. The association of abdominal wall defect and limb anomaly like kyphoscoliosis imply the diagnosis of limb body wall complex. The content, cord attachment and associated anomalies help in differentiating this between omphalocele and gastroschisis. See table below:

Cloacal / Bladder Exstrophy

Bladder Exstrophy: Exstrophic anomalies are a group of disorders derived by maldevelopment of caudal folds of the anterior abdominal wall. In bladder exstrophy, the anterior wall of the bladder is absent and the posterior wall is exposed. Normally the uro rectal septum divides the cloaca into anterior uro genital sinus and posterior hind gut sinus and the mesenchyme proliferates to form the lower abdomen. Cloacal membrane becomes the anterior wall of the bladder. When mesenchyme does not proliferate regression of cloacal membrane results in direct exposure of the post bladder wall.

Diagnosis& Management: Persistent non visualization of the of the bladder in the lower part of the abdomen and low placed umbilicus and protrusion of post vesical wall are suggestive of bladder exstrophy. Incomplete descent of testes, short penis, epispadias in males and cleft clitoris in females may also be seen. Bladder exstrophy can be surgically repaired but this in general has a long term morbidity and also high mortality.

Cloacal exstrophy: In this anomaly the uro rectal septum does not divide the anterior and posterior parts. Cloacal membrane forms the anterior part of the abdominal wall instead of the mesenchyme. The bladder and the rectum are not separated. When the cloacal membrane regresses both the bowel and the bladder get exposed.

Diagnosis& Management: If the ventral wall defect is seen with bladder exstrophy, the diagnosis would be cloacal exstrophy. Unlike bladder exstrophy, this is generally

associated with renal abnormalities, omphalocele, spinal defects, limb defects and has a elephant trunk appearance due to prolapse of the ileum. Prognosis for cloacal exstrophy is not good. Surgical correction involves many stages and post surgical complications include, bladder incontinence, infertility, uterine prolapse, ureteric infection and malignancy.

Discussion:

• Of the abdominal wall defects discussed, which are the ones that warrant karyotyping?

Conditions like omphalocele,cloacal/bladder exstrophy warrant karyotyping,. Others such as gastroschisis, limb body wall complex, body stalk anomaly are sporadic anomalies and hence do not require karyotyping.

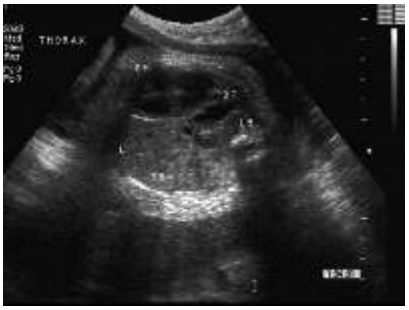
• How does umbilical hernia differ from omphalocele?

Umbilical hernia is the defect of the abdominal layers and no treatment is needed for it. Umbilical hernia is innocuous unless it is huge where as omphalocele is a major problem and it needs post natal surgical intervention.

MISPLACED ORGANS IN THE GIT:- Dr. Susila Vavilala

Dr.Susila said that to maintain harmony in life everything should take its position in the right place. So the word "Situs" denotes the placement of an organ in the body. The right and left sides of our body are identical at a glance. Internally some of the internal organs are displaced to one side. In a normal fetus the stomach and heart are in the left side. Establishment of normal situs is essential in obstetric scans. Asymmetric expressions of genes lead to malpositioning of organs.

Significance of situs: Normal visceral situs establishes normal anatomy. The outcome in situs inversus totalis is usually normal. Situs ambiguous is invariably associated with cardiac anomalies and hence it mandates a thorough anatomic survey and fetal echo. In the process of development, the right and the left umbilical vein join the corresponding portal veins and drain in to inferior vena cava. If left umbilical vein is left behind, it is normal. If right umbilical vein is left behind , then it is called persistent right umbilical vein. It is a normal variant and this finding mandates a complete fetal survey and fetal echocardiogram. Congenital diaphragmatic hernia: Sometimes, some of the abdominal organs get displaced due to a defect in the diaphragm called congenital diaphragmatic hernia. The stomach, intestines, spleen and liver herniated in to the chest depending on the site of the defect in the diaphragm. Cardio-mediastinal shift is the signpost for diagnosing this condition. A right sided congenital diaphragmatic hernia should always be included in the differential diagnosis of right sided chest mass. Perinatal mortality depends on the size of the hernia, gestational age, associated anomalies or chromosomal anomalies.



Prognosis is good if the lung head ratio is greater than. Liver as content has poor prognosis and invasive fetal therapy has not proved successful. If prognosis is good, the baby has to be delivered in a tertiary centre and surgical management is required further.

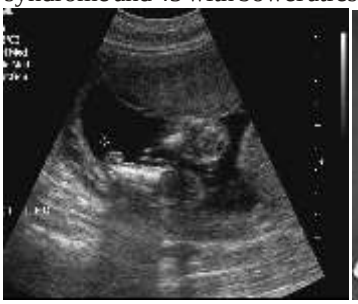
Discussion:

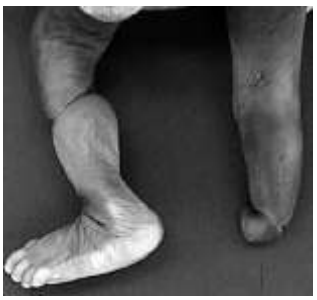
• How does eventration of diaphragm differ from diaphragmatic hernia? Does it warrant karyotyping?

Eventration is thinning of the diaphragm. If it is established without doubt, karyotyping is not necessary as it is not associated with chromosomal anomalies unlike diaphragmatic hernia where it is mandatory.

SYNDROMES ASSOCIATED WITH GIT - Dr. Radhika Ramesh

Dr. Radhika's presentation about the syndromes associated with GIT anomalies helped the audience appreciate the wide spectrum of syndromes associated with the gut. She explained the importance of understanding the most common syndromes and associations to make precise risk predictions. There are about 650 reported syndromes of the GIT. Fetal syndromes can be fixed by analyzing the spectrum of anomalies. GIT anomalies may be associated with 1) Cranio facial anomalies, 2) Cardiac anomalies 3) Renal anomalies or 4) multiple markers. "Syndrome" is a Greek word which means "things that run together". They are a pattern of multiple anomalies thought to be pathogenetically related and not to represent a sequence. It is necessary to have a good understanding of the following points such as the developmental mechanism of the embryo-- structural derangements that happen in similar tissues (for eg ectoderm)/ areas, and those systems that share the same genes (for eg bone & limbs), the time of insult in embryogenesis and whether it is a known association or sequence. The causes underlying the syndromes can be 1) chromosomal, 2) single 3) terotogenic, 4) vascular disruption or 5) sporadic. She enumerated a few common anomalies of GIT and its vast associations and syndromes with challenging case scenarios. To put it precisely she explained that 72 syndromes are reported to be associated with esophageal atresia, 42 syndromes with duodenal atresia, 94 syndromes with diaphragmatic hernia like Fryn syndrome, Edward syndrome, Amniotic band syndrome and Facio auriculo vertebral dysplasia, 69 syndromes with amniotic band syndrome and 45 with bowel atresia.





Why to fix a syndrome?

Syndromic diagnosis facilitates better understanding of a specific pattern of multi system malformations, mode of inheritance, recurrence risk prediction and genetic counseling. It also serves as a ready algorithm for the sonologist while scanning in the subsequent gestations.

How to fix a diagnosis?

Professionals need to be aware of associations of certain multi system anomalies. When fetal anomalies are seen, postnatal evaluation is mandatory with a multidisciplinary approach. POSSUM-Australia, OMIM (Online Mendelian inheritance in man), London Dysmorphology database are some of the resources where one can look for reference.

Discussion:

•While giving USG report for a fetus with anomalies, is it necessary to name it as a syndrome?

That is the job of the genetic counselor. In the absence of counseling services in a set up, all sonographic findings could be listed out in the report for immediate obstetric management and later it could be assigned a syndromic diagnosis if need be.

GENETIC COUNSELING FOR GIT ANOMALIES - Dr. Susila Vavilala

Pregnancy with a fetal malformation is an extremely stressful condition. The couple need a lot of emotional support throughout pregnancy and after delivery.

GIT anomalies could be broadly classified in to 1) Ventral wall defects, 2) Bowel disorders 3) Cystic leisions and 4) Associations and syndromes suitable protocols for genetic counseling could be formulated. Genetic counseling is a communication of information and advice about an inherited disorder. A secure diagnosis forms the basis of counseling. Family pedigree, index child work up and autopsy details in terminated pregnancy facilitate effective counseling of the couple concerned. The whole process should be non directive. It is ideal to counsel before conception and follow through pregnancy and after the birth of the baby.

Genetic counseling requires a multidisciplinary approach involving a sonologist, fetal medicine specialist,a dysmorphologist, an obstetrician and a neonatologist.

EARLY DETECTION OF CORRECTABLE CONGENITAL GIT ANOMALIES - Dr. S. Balagopal.

Dr. Balagopal started with a positive note saying that his job as a surgeon is easier when compared to an obstetrician/ sonologist as he sees live babies and not through the mother. Surgeon has more time for decision making he added. His presentation was backed up by beautiful illustrations for better understanding.

Clinical Scenario:

He listed out the most commonly seen GIT problems that could be surgically managed.

Those were - Tracheo esophageal atresia, Pyloric stenosis,duodenal atresia/stenosis, Annular pancreas, Malrotation, Intestinal Atresia/stenosis, Meconium ileus/ peritonitis, Duplications, Hirschsprung disease and Anorectal anomalies.

• Most of these present as polyhydramnios antenatally. Postnatally detailed clinical evaluation is mandatory to understand the family history, drug intake and infections during pregnancy, associated problems and clinical manifestation such as vomiting to narrow down the diagnosis.

• If there is a green bilious aspirate in the nasogastric tube it is suggestive of intestinal obstruction. When there is non bilious vomiting and only non swallowed saliva and froth are seen, it is suggestive of tracheo esophageal atresia or pyloric stenosis. In tracheo esophageal fistula, air is pushed down causing abdominal distension.

• Abdominal distension above the umbilical cord is suggestive of duodenal atresia /malrotation of the proximal bowel. Visible peristalsis and tenderness of the abdomen suggest bowel compromise and lower intestinal obstruction. Feeds need to be curtailed in these situations.

• Non passage of meconium is also to be considered when there is distension. If there is only mucous plug, a rectal wash would solve the problem.

• Haemetemesis and haematochesis are dreadful signs and suggestive of midgut volvulus. Sometimes babies swallow the maternal blood during delivery and this has to be ruled out in such situation.

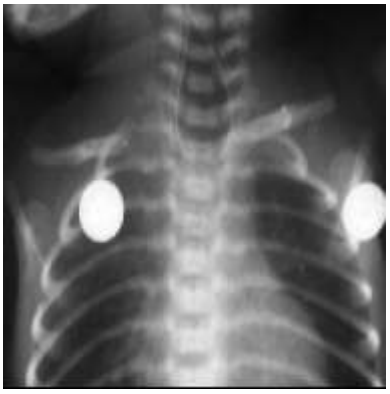
Investigations:

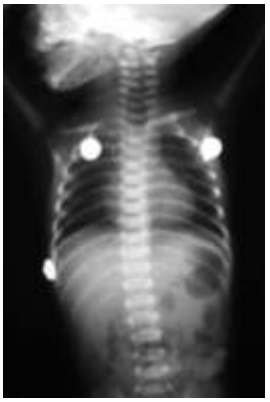
On the whole, investigations such as haematological (CBC), serum chemistry, imaging of the abdomen by USG/CT/MRI with contrast, and radiograph complement accurate diagnosis and management. The surgeon dealt in detail about the surgical management of some of the common GIT anomalies.

Esophageal atresia:

It is important to find out first whether there is Tracheo esophageal atresia with or without fistula.Tracheo esophageal artresia with fistula is more common in our scenario. In this condition saliva pools in the trachea causing respiratory distress. Diagnosis is made when there is non passage of the nasogastric(NG) tube into the stomach. X ray of the chest with the NG tube will show the coiling. This reveals the level of obstruction. He stressed the need for careful handling to prevent perforations while inserting the tube. For isolated esophageal atresias, pre operative ventilation is not necessary.

Primary repair is done by right thoracotomy and by ligation and division of fistula of the tube. Post operatively the baby needs adequate nursing care and ventilation. The ultimate goal of the surgery should be to put the native esophagus together by anastomoses, though quite a few problems like regurgitation will have to be managed postoperatively. This is possible as long as the baby is alive and responding to treatment. Though replacement option is mentioned in text books, it is not so practical to follow it. Prognostication in these surgeries is a difficult task whatever may be the risk factors involved.





Duodenal atresia:

Duodenal atresia as discussed already is associated with Down syndrome and sometimes with annular pancreas. The baby presents here with bilious vomiting and upper abdominal distension. X ray of the abdomen or contrast study shows double bubble-a gastric and duodenal sign confirming the

diagnosis. Treatment consists of anastomoses by perfoming Duodeno duodenostomy. Even when there is annular pancreas this is done side to side not touching the pancreas. Sometimes a bypass is done with jejunum by duodeno duodenostomy procedure. Prognostication is again difficult as babies with duodenal atresia are generally small babies.

Malrotation:

Malrotation as already discussed is due to an error in rotation and fixation of the bowel. The small bowel and the duodenum do not go across the midline and come straight down causing problem. Line of fixation should be to the left of spine and to the iliac fossa on to the right. When this does not happen, there is loose lying mesentery and narrow pedicle.The reason for non fixation of midgut during the fetal which causes this phenomenon is not clear. The surgeon attributed the cause to one's destiny.The Ladds bands fix the caecum to the abdomen compressing the duodenum causing bilious vomiting. Widely placed bowel in the narrow pedicle causes the twist. Diagnosis is by X ray, USG, and contrast study. X ray shows some gas filled loops and double bubble sign. USG reveals the alteration in the orientation of superior mesenteric artery and the superior mesenteric vein. The degree of rotation should also be understood which may vary from 90 - 180 degrees. If it is 360 degrees, then it may mimic a normal pattern. Contrast study and barium enema to locate the cecum is also helpful in fixing the diagnosis. Clinically the child presents this as bilious vomiting, duodenal obstruction and bleeding in NG tube. The treatment is by surgically reducing the volvulus, division of Ladds bands, widening of the mesenteric pedicle and appendicectomy.

Jejuno ileal atresia: The stenosis here is caused due to an ischemic insult. There are 4 types of this atresia

Type I - Mesentery maintains continuity with obstructing membrane

Type II- Mesentery maintains continuity but not the bowel

Type III - Mesentery and bowel have defects

Type IV - Multiple atresias present in the bowel. Antenatally these present as polyhydramnios. The baby has bilious vomiting, non passage of meconium and abdominal distension. X ray shows gas filled dilated loops. On rectal wash some meconium along with large amount of mucus would come out. Correction is done by excision of maximum dilated loops and anastomosing the rest of the bowel. Ganglionation here will not be a problem as ischemic insult occurs much later and by then ganglion cells would have migrated.

Meconium ileus:

Pellets obstructing the bowel passage is seen in meconium disease. This is usually associated with cystic fibrosis. Meconium here is sticky and the clinical manifestation is similar to that of atresias. X ray shows soap bubble appearance due to the presence of meconium with air. Fluid level is absent in meconium ileus unlike atresias. Rectal wash easily solve the mucus plugs. However it is mandatory to rule out Hirschsprung disease and Cystic fibrosis while treating this condition. This is usually found in diabetic mothers and in babies having small left colon syndrome.

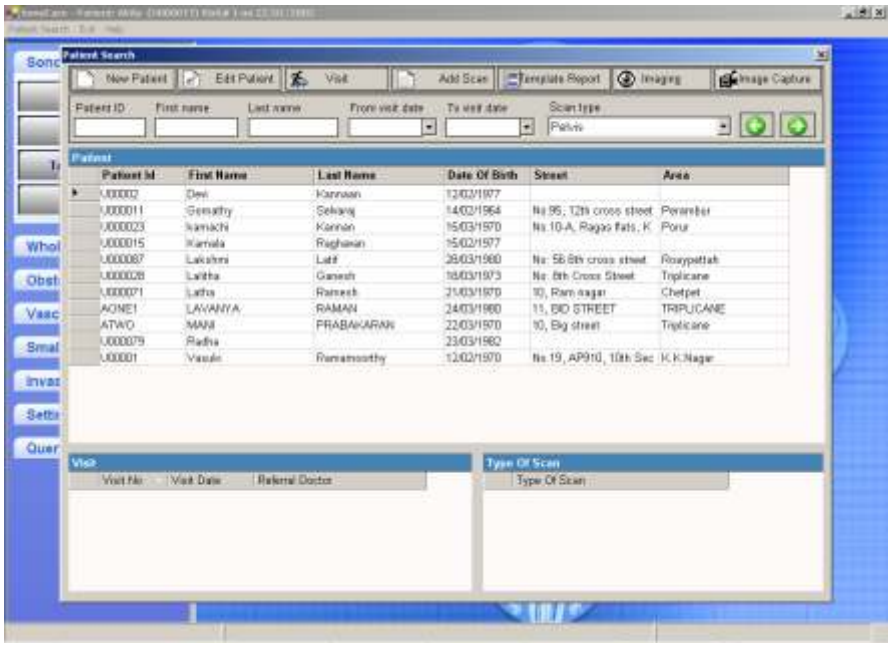
Hirschsprung disease: Aganglionosis of colon usually involving the recto sigmoid region causes delayed and prolonged passage of meconium in babies with Hirschsprung disease. Abdominal distension can be rectified by giving a rectal wash. Further investigations can be done with anal manometry, rectal biopsy by suction or open method and contrast enema. Giving enema in newborns is difficult unless there is a long segment involved. Management is through diversion colostomy in the new born period and later the second surgery is done by 9 months of age. Pull through surgery creates heavy morbidity in the new born and not opted these days. The surgeon spoke briefly about the surgical management possible for exomphalos, gastroschisis and duplication cyst of the terminal ileum. He said though success depends on a number of factors in all these cases, with careful planning they can be managed.Ano rectal malformations: Anorectal malformations include absent anus/ectopic anus/with or without fistula. If there is a fistula meconium may be present in urine. Perineal examination is essential depending on the gender of the babies to know the presence of orifices. This is usually present in association with Vacterl. Investigation is by X- ray after birth. Pubococcygeal(PC) line is drawn to find out the level of anomaly. If it has gone past the sphincters, it is a low anomaly and anoplasty can be done. For high anomaly, diversion colostomy is done initially followed by pull through surgery later.

Dr. Balagopal concluded saying that it is very difficult for a surgeon to suggest termination even for fetuses requiring complex multistage surgeries of the gut. Though all paediatric surgeries involve greater risks, many things such as the money involved, family situation have to be considered by individually offering surgical options rather than abiding by fixed norms to solve the problem.

An appeal

Help a national cause Join the Birth defects registry. If you are already a member of the registry, please motivate a friend to become a member of the registry. If you are not a member kindly contact us. Let us work together to build a healthier nation.

This news letter is available online at http://www.mediscansystems.org. Issued four times in a year - January, April, July and October. Published by Fetal Care Research Founation, 203, Avvai Shanmugam Salai, Royapettah, Chennai - 600 014. For Private circulation. Printed at The Print Shoppie (Print Supplies), Ayanavaram, Chennai - 600 023.



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