



B D R News

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

Volume 8

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Proceedings of the birth defects registry meeting held on 25.10.2008

The fourth & final BDR meeting of the current year was held on 25th October 2008 at the MediScan premises, Chennai.

Dr. S. Suresh extended a warm welcome to the audience. He said that FOGSI BDR launched in the current year is gaining momentum & a few societies under the Federation of Obstetric & Gynaecological Society of India are making serious efforts to form registries in their respective regions. He mentioned that he would be addressing the FOGSIANS at the annual meet in January next year to motivate them to actively get involved in this national project. He hoped to access a large data through FOGSI BDR network with a wider coverage of births in India. He appreciated the efforts of FOGSI Navi Mumbai to commission a registry first with a team of more than 40 hospitals. Dr. Anu Vij of Anurag Hospital would be steering this as the nodal member of FOGSI Navi Mumbai BDR.

Talking about the importance of 11-14 scan during pregnancy, he once again mentioned about the initiative of the Down syndrome Association of Tamilnadu to facilitate antenatal invasive procedures & diagnostic tests free of cost for the under privileged pregnant women. He appealed to members to avail this facility for deserving cases found to be positive in I / II Trimester screening tests. He also requested them to provide more case samples of Neural Tube defects (NTD) for the Government sponsored DBT project to understand the etiology of NTD & the efficacy of periconceptional Folic acid and Vitamin B₁₂ supplementation. The results of the preliminary study completed so far have revealed interesting facts. We would be getting a protocol for prevention of the most common birth defects in India at the end of the study in another 2 years he added.

He also spoke about Fetal Care Research Foundation (FCRF), a not for profit unit of MediScan. It was founded for the cause of birth defects in India. It is facilitating therapeutic treatment for a few Lysosomal Storage Disorders (LSD) such as Mucopolysaccharidoses, Gaucher, Pompe & Fabry diseases. Genzyme Corporation, U.S.A has formed an Indian Medical Advisory Board (IMAB) to offer Enzyme Replacement Therapy (ERT) free of cost for children afflicted with any one of the disease mentioned above in India. Dr. Sujatha Jagadeesh (Dysmorphologist, BDRI) is a member of the advisory panel & she could sponsor ERT for the affected with diagnostic confirmation. Therapeutic care for rare genetic disorders is sponsored in the West as it involves exorbitant cost throughout life. Dr. Suresh said that previously our approach to such diseases used to stop with diagnosis & providing temporary treatment to alleviate the symptoms. He appealed to members to refer relevant cases for treatment as now it

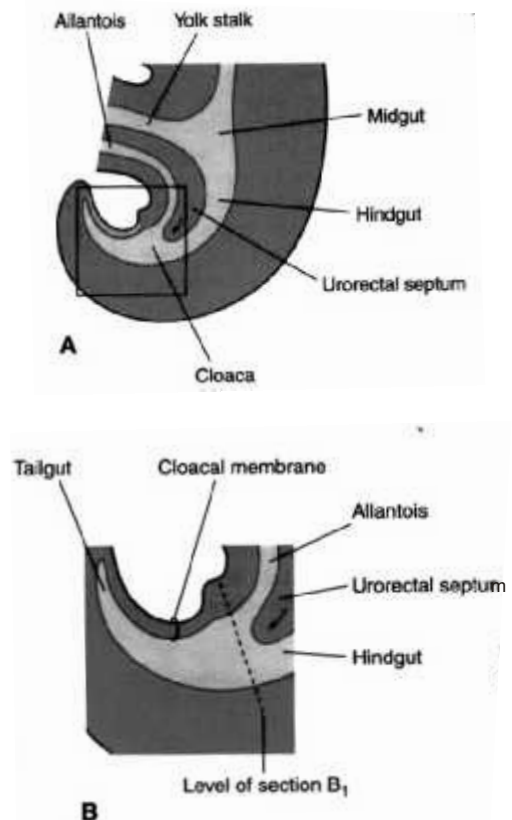
is made available free of cost to provide better quality life to patients. He announced the CME session for the day on Anorectal & Cloacal anomalies & introduced the speakers. The excerpts of the presentations are given below:

Ultrasound imaging and diagnosis of Cloacal & Anorectal anomalies

Dr. Aparna Kulkarni, Fellow, Fetal medicine, MediScan

Dr. Aparna Kulkarni commenced the session with Cloacal anomalies. Cloaca is the sewer or common excretory outflow system of the human body. Cloacal abnormalities seen in antenatal period may be of **3 types** namely

1. Cloacal Dysgenesis sequence 2. Cloacal Exstrophy sequence commonly represented as OEIS complex. (Omphalocele, Exstrophy of bladder, Imperforate anus, Skeletal defect) & 3. Persistent Cloaca/urogenital sinus. These are **very rare** and are mostly associated with other structural anomalies. **Embryology** of Cloaca: Between 6th & 7th week of gestation, the urorectal membrane of mesenchymal origin forms between the allantois in the anterior part of the embryo & the hind gut in the posterior position. The Cloacal membrane as seen below covers



the anterior part during this period, fuses with the uro rectal septum forming urogenital sinus anteriorly, rectum & anal canal posteriorly. During the 8th week the Cloacal memberane ruptures which is a normal phenomenon thereby forming three clear-cut outflow passages. Before the 8th week, the Cloaca remains as the common excretory passage in the human embryo. If the cloacal membrane 1. does not rupture or 2. ruptures before the 8th week or the uro genital membrane does not form, the fetus has Cloacal defects.

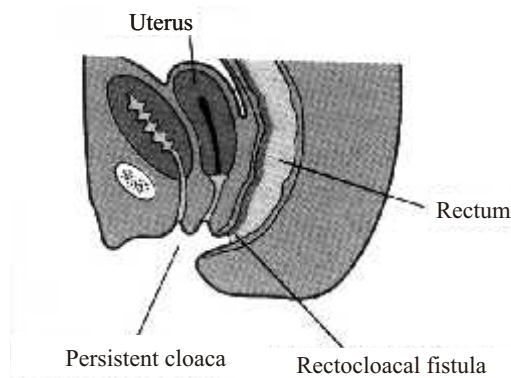
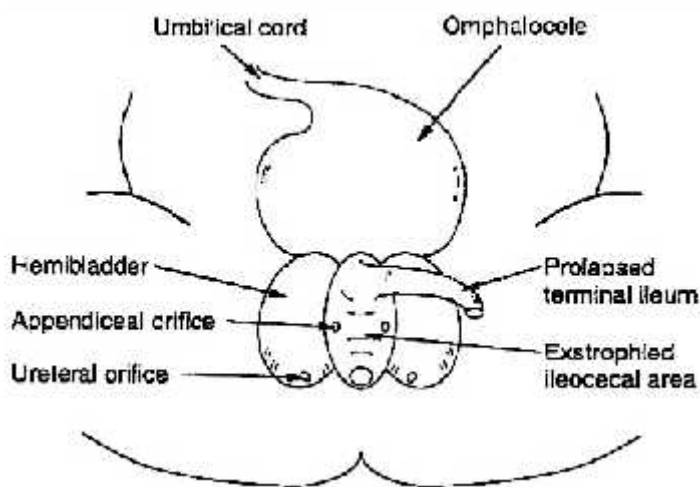
In **Cloacal dysgenesis** sequence there is absence of anal, urinary & genital orifices with a smooth perineum. Other commonly associated defects in this sequence are absent / dysplastic kidneys, hydro ureters, mega cystis & pulmonary hypoplasia. Ultrasound features include dilated fluid filled colon, enterolithiasis, cystic structure in the pelvis.



Persistent Cloaca - results due to failure of formation of urorectal septum.



Cloacal exstrophy, which is also known as Vesico - Intestinal fissure is also a rare complex congenital anomaly with an incidence ranging from 1/200,000 - 1/400,000. This results from migration failure of the lateral mesodermal folds of the infra-umbilical anterior abdominal wall and rupture of the resulting enlarged, persistent Cloacal membrane before the 8th week of gestation. The **characteristic features of Ultrasonogram (USG) in Cloacal exstrophy** are: lower anterior abdominal wall defect with omphalocele or cystic anterior abdominal wall structure in contact with amniotic fluid, persistent non visualization of bladder, skeletal abnormalities & abnormal genitalia. The **anatomical features of Cloacal exstrophy** would include, omphalocele, imperforate anus, hemi bladder with ureteric opening on either side, open caecum in between the bladder, blind ending ileum hanging down in the pelvis from the caecum, widely separated pubic bones, bifid phallus, bifid clitoris & double vagina.



The urorectal, genital & intestinal tract converge into common outflow - cloaca & there is no differentiation between the urogenital sinus & the hind gut. There could be one or two perineal opening in such cases. This defect is seen more in females. The **USG features** of this anomaly include: distention of vagina, rectum, bilateral hydro nephrosis with cystic kidney & oligo hydramnios.



It is commonly associated with IUGR, vertebral, cardiac, renal & intestinal defects. The prognosis of fetus with Cloacal malformation is poor due its association with other structural defects & oligohydramnios.

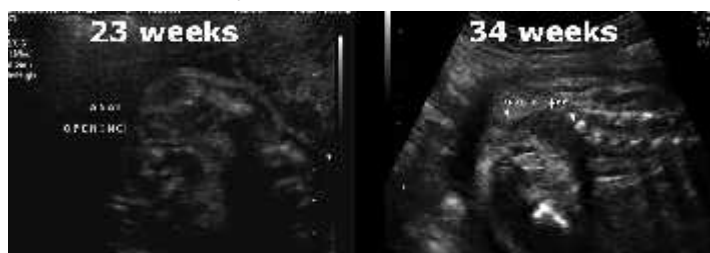
Anorectal malformations (ARM) are malformations causing distal obstruction of the Gastro intestinal system presenting as imperforate anus or anal atresia or ano-rectal malformation with or without fistula. They result from failure of rupture of Cloacal membrane. The upper part of the anal canal- hind gut is endodermal in origin & the lower part of the anal Canal is ectodermal in origin. The incidence of ARM is very rare..

They may be part of chromosomal anomalies such as Trisomy 18 & 21 and non chromosomal anomalies such as Caudal regression syndrome, VACTERL & Sirenomelia.

Anorectal anomalies may be **classified** according to embryological origin such as **external malformation** caused by the abnormal development & fusion of the external perineal layers- Eg: Imperforate anus with or without fistula & **internal malformation** caused by anomalous partition of the Cloaca by the urorectal septum- Eg: rectal atresia with or without fistula or mixed malformation at all possible sites of ectopic anus. According to **anatomical level** they are **classified** either as 1) High (supra-levator lesions) anorectal anomalies which are more common and are associated with fistulas & genito urinary malformations 2) Low (infra-levator lesions) anorectal anomaly. However it is not feasible to predict the level of lesion by ultrasonography.

USG imaging of these defects is difficult in first /second trimester unless there is any obstruction. The sensitivity of USG to diagnose isolated anorectal defects antenatally is very low. (8% Bruce et al, Pediatric Surgery International Vol 10(1) Jan 1995). **Ultra sound** imaging provides some **clues** in III trimester to suspect anorectal defects. They may be 1) over distension of the rectum and to a lesser extent of the sigmoid 2) the diameter of the distal rectal pouch > diameter of the full bladder 3) hyper echoic / calcified meconium pellets in the dilated rectal pouch & 4) liquor may be normal, reduced in case of anorectal atresia with recto vesical fistula, increased in case of other associated anomalies. Imaging of prominent bowel loops during first trimester which are not to be visualized are suggestive of anorectal malformation. Apart from this decreased levels of liquor, MSAFP & microvillar enzymes also provide a clue for diagnosis of these defects. One should think of colo-vesical/recto vesical fistula if he/she sees echogenic debris in the bladder and or fluid in the rectum / sigmoid colon

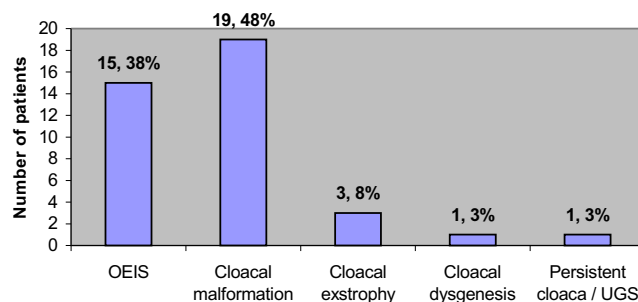
USG of normal anal canal & rectum



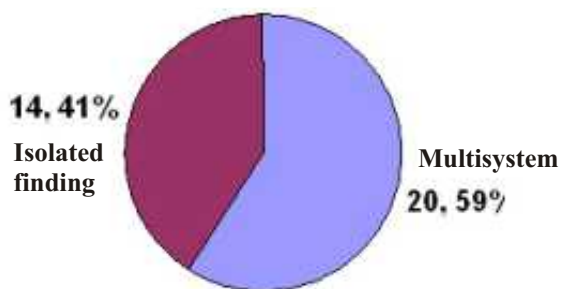
USG showing echogenic debris in the bladder & fluid in the rectum

As for **associated anomalies** are concerned, the most common are urogenital (70-90%) followed by skeletal/spinal anomalies (30%), GIT (TEF) (10%), cardiac (5%) & central nervous system (2-3%). **Differential diagnosis** for anorectal defects would be various presentations of small bowel dilatation, persistent cloaca & Hirschsprung disease. The **prognosis** of fetus with anorectal anomalies is poor due to associated malformations. Bladder & bowel incontinence are commonly reported after surgical repair. She ended her presentation by reporting the data collected between January 2001-October 2008 at Mediscan on Cloacal & Anorectal anomalies. 39 cases of cloacal anomalies and 34 cases of ARM were retrospectively studied.

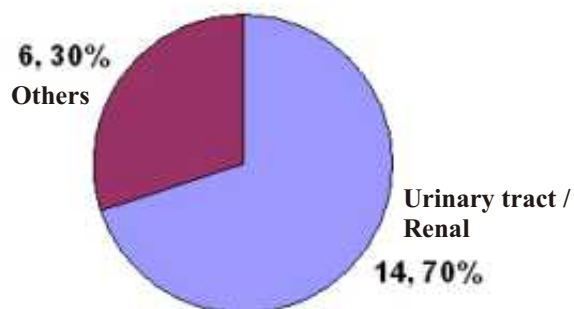
Cloacal defects (n=39)



Anorectal malformations (n=34)



Multisystem abnormalities



Urological management issues in Anorectal and Cloacal anomalies in children

Dr.V.Sripathi, Consultant Pediatric Urologist, Chennai

At the outset, Dr.V.Sripathy was reminiscing about the International Conference where he presented a paper on the same topic in Germany a few years ago. He won great appreciations from leading expert in the field - Prof. Alberto Pena for his presentation. He thanked Dr. Suresh for providing valuable, original ultra sound data that enabled him to present the paper. He also acknowledged BDRI for providing a

baseline data on birth defects that gave a glimpse of the incidence of anorectal / cloacal malformations in India. He stated that his presentation on Ultrasound Diagnosis of Anorectal anomalies authored by himself along with Dr. Suresh & Dr. Sugirtha also got published by March of Dimes Birth Defects Foundation that set the cornerstone for a decade.

Abnormalities of the urinary tract constitute 40% of all Anorectal anomalies while genital tract anomalies exist in 10%. It is therefore essential to screen for urinary and genital anomalies in all children with anorectal anomalies and plan appropriate treatment.

If the child with an anorectal anomaly has a fistula with the urinary tract, the incidence of urinary anomaly increases in proportion to the level of the fistula as follows:

Level of fistula	Incidence of genitourinary anomalies
In males:	
Fistula to the bulbar urethra	25%
Fistula to the prostatic urethra	66%
Fistula to the bladder	92%
In females:	
Vestibular fistula	30%
Cloaca	88%
In both sexes	
No fistula	25%
Perineal opening	Nil

Kidney Anomalies in children with anorectal anomalies:

These may be in the form of a pelvic kidney, horseshoe kidney or crossed fused ectopia. These anomalies predispose to urinary infection and vesicoureteric reflux and need followup. Renal duplications may be a cause of wetting if the upper moiety ureter is ectopic. If a hydronephrosis is detected careful follow up is needed as approximately 25% will need surgical correction. Renal dysplasia (including multicystic dysplasia) and renal agenesis are also often encountered. Children with anorectal anomalies with single kidney status need to be carefully monitored for renal failure.

Even if both kidneys are present 6% of children with high anomalies and 1% of children with low anomalies suffer from Chronic Renal Failure with a Glomerular Filtration Rate of <80mls/min. However due to the multiple surgeries done in these children, dialysis and renal transplantation are difficult in this group

Ureteric Anomalies in children with anorectal anomalies:

Vesico ureteric reflux is seen in 20 - 30% of these children. Obstructive megaureters and ectopic ureters with or without duplications are also seen.

Genital Anomalies in children with anorectal anomalies:

More than half of these children have some form of genital anomalies. In males hypospadias, bifid scrotum and penoscrotal transposition are seen while in females septation of the vagina, cervix and uterus are often encountered. Muellierian abnormalities

are also very common. Hamartomas and hemangiomas of the labia and perineum are often seen in female children.

Functional Problems of the Urinary Tract in children with anorectal anomalies:

If a child with an anorectal anomaly presents with Lower Urinary Tract Symptoms (LUTS) in the presence of normal kidneys, the spine should be assessed. 88% of these children have problems either with the bony sacrum, spinal cord or filum terminale. The incidence of such problems is more in high than in low anomalies.

Investigation of the urinary tract in a child with anorectal anomaly:

These include:

- Renal ultrasound
- Micturating Cystourethrogram to assess bladder contour (neurogenic or not), presence or absence of vesicoureteric reflux, and presence of fistula with the urinary tract.
- Isotope renogram to assess the function of the kidneys.
- Spinal Status assessment This includes vertebral x-ray (AP and Lateral views), ultrasound assessment of the spinal cord (in children < 3 months of age) and MRI of the cord (in children > 3 months of age)
- Urodynamic assessment of the urinary tract is mandatory in all children with high and intermediate type anomalies.

Investigations in children with cloacal anomalies

- Renal functional assessment on a periodic basis to document renal function deterioration.
- Mandatory Spinal Status assessment.
- Mandatory preoperative urodynamic study.
- Assessment of length of common channel (< 3cms or >3cms) by contrast study and scopy.



*Perineal appearance of cloacal anomaly
The urinary, genital & anal tracts opening in one short channel (<3cm) in the vulva*

Long Term follow up study to assess problems related to the genital tract.

Urinary Incontinence in children with Anorectal anomalies:

Risto Rintala in an assessment of 35 year old adults with anorectal anomalies found 33% of high and intermediate anomalies to be incontinent of urine. In cloacas especially in those with a common channel of >3cms, incontinence is even higher (50-70%). The cause for incontinence is two fold:

- Denervation of the pelvic autonomic plexus during surgery as the plexus is abnormally situated (close to fistula site).
- Severe constipation, which presses on the bladder promoting incontinence.

Management of incontinence:

Two tests are necessary:

- Postvoid residue assessment
- Urodynamic testing

If the post void residue is more than 10% of the expected bladder capacity for age then Clean Intermittent Catheterisation (CIC) is commenced and the bladder is emptied at least 4 - 6 times a day. CIC eliminates post void residue, urinary infection and protects the kidneys. Most children with anorectal anomalies have sensate urethras and hence will not allow the urethra to be used for passage of a feeding tube.

In such cases Mitrofanoff Appendicovesicostomy is performed. In this procedure the appendix is separated with its blood supply from the cecum and the tip is implanted in the bladder. The other end is brought out on the skin of the abdominal wall. The lumen of the appendix is used as a pain free channel for passage of a tube into the bladder. The appendix being well vascularised grows with the child.



Appendix on its blood supply separated from the Cecum prior to reimplanting in the bladder Mitrofanoff Appendicovesicostomy

Tip of Appendix implanter in bladder of Cecal end brought out in belly button for Clean Intermittent Catheterisation (CIC)



If the bladder shows high pressures on filling in urodynamic testing, enlargement of bladder is done to enhance its capacity by doing an augmentation cystoplasty. This involves putting a patch of the colon or small bowel on the bladder to enlarge its storage capacity and to reduce its pressure. Refer fig below

Augmentation Cystoplasty completed in a child with anorectal anomaly with high pressure bladder. The lower part is the filled bladder and the distended 'bishops cap' represents the small bowel sutured to the bladder as a cup.



Urethral problems after anorectal anomaly surgery:

These include:

- Urethral injury due to ligation of fistula close to urethra.
- A fistula ligated far away from the urethra can dilate to become a diverticulum, which can harbor stones and cause recurrent urinary infections.
- Children with anorectal anomalies can develop recurrent Epididymitis due to an ectopic vas.
- Children with prostatic and vesical fistulae can develop ejaculatory problems and have poor sperm counts in adult life.

Recent Advances

The advent of Laparoscopy assisted Anorectal Plasty in which minimal dissection is done to ligate the fistula and to bring down the bowel is believed to reduce the postoperative problems cited above to a large extent. However long term follow-up is needed to substantiate these claims.



Marking the sites of port placement for Laparoscopy Assisted Anorectoplasty. The sigmoid colostomy is seen.

Anorectal Malformation - Surgeon's perspective

Dr. S. Balagopal, Consultant Paediatric Surgeon, SRMC Hospital

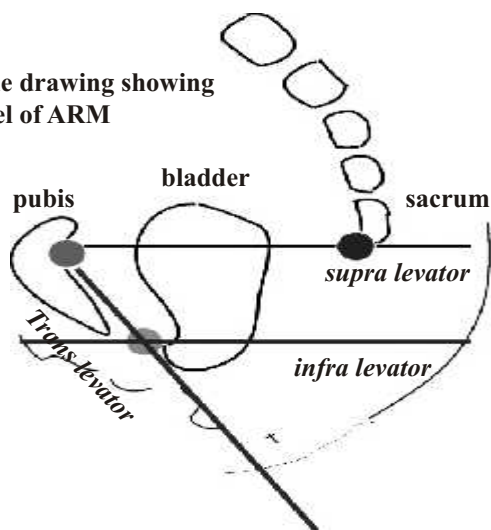
Dr. Balagopal stated that parents with children requiring surgical repairs expect total normalcy in them after the surgery. It is not always possible to achieve complete success in cases like ano rectal malformations. He presented an outline of the topic before speaking on the surgeon's perspective of anorectal malformations (ARM). They tend to be high & severe lesions with Recto urethral anomaly defect being the commonest in males. Ano vestibular lesions are the commonest among females. Their etiology is mostly

unknown though familial presentation has been reported. Siblings are not usually affected & male predominance postulates sex linked inheritance. Postnatally while examining the baby with anorectal anomaly, the surgeon has to exercise great patience, observe the developments (Meconium passage) & then proceed with necessary diagnostic investigations. In antenatally diagnosed cases, the families would be anxious to get the repair done at the earliest. The common clinical features of anorectal defects are:

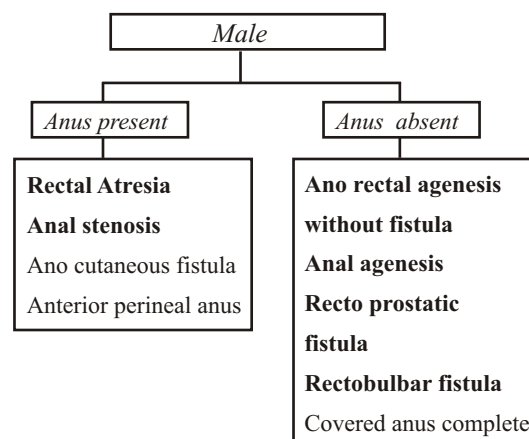
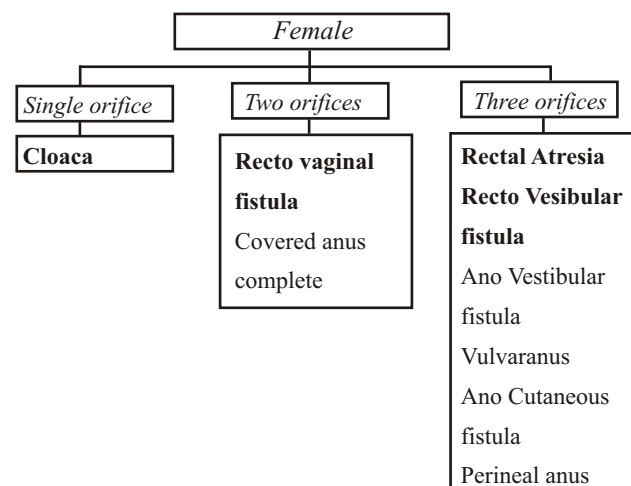
- Perineal fold or raphe related or unrelated to anal orifice
- Bucket handle opening
- White epithelial folds
- Meconium pearls or staining in the tip of urethra
- Cry impulse
- Flat perineum
- Sacrum & coccyx dysgenesis
- Perineal sensation & twitching
- Associated signs of low intestinal obstruction, associated discharge via urethra (post voiding urethral compression over a gauze) & associated cleft scrotum, hypospadias,
- Undescended testes, wall defects



Line drawing showing level of ARM



The following flow chart provides guidance about the level of lesions in both sexes during clinical examination.



Please note:

1. Bold words denote high and thin words denote low anomalies.
2. In the male if anal orifice is not found it is usually a high anomalies except for a covered anus complete.
3. In the female if anal orifice is found it is usually a low anomalies except for rectal atresia.

He explained that the four group of muscles namely the levator ani complex, internal, external sphincter muscles & the pubo rectalis play an important role in planning a corrective surgery for ano rectal malformations. The success of the surgery is related to muscles already present & preservation of these muscles, which determine the normal bowel function after surgery. If the rectum is already formed & it has passed through the levator ani complex, procedures are done from the base. The surgeon's opinion was that single stage perineal pull through surgery has a higher morbidity over spaced abdominal colostomy. Laproscopic interventions have its own variable outcome in this picture. He presented various types of classifications including the International classification (refer table in the next page) of ARM depending on their location through the levator ani complex. Low ARM have better surgical outcome. The reason being that they have already passed through the levator ani /sphincter complex only less damage can happen to nerves & muscles in surgical procedure.

International Classification – 1970.		
	Male	Female
High Deformities (Supra Levator)		
Ano Rectal Agenesis	Without Fistula	Without Fistula
	With Fistula Recto Vesical Recto Urethral	With Fistula Recto Vesical Recto Cloacal Recto Vaginal(High)
Rectal Atresia	Rectal Atresia	Rectal Atresia
Intermediate (Trans Levator)		
Anal Agenesis	Without Fistula	Without Fistula
	With Fistula Recto Bulbar	With Fistula Recto Vaginal (Low) Recto Vestibular
Ano Rectal Stenosis	Ano Rectal Stenosis	Ano Rectal Stenosis
Low (Infra Levator)		
Normal Anal Site	Covered Anus Complete Anal Stenosis	Covered Anus Complete Anal Stenosis
At Perineal site	Anterior Perineal anus Ano Cutaneous Fistula Incomplete covered anus Perineal Canal & Groove	Anterior Perineal anus Ano Cutaneous Fistula Incomplete covered anus Perineal Canal & Groove
At Vulvar Site		Vulvar anus Ano Vulvar fistula Ano Vestibular fistula

A few syndromes such as Cat Eye syndrome, Towne's syndrome & FG syndrome were mentioned. In general 9% of GUT anomalies are associated with low defects & 20-60% with high defects. 12-25% of CVS anomalies such as Tetralogy of Fallot, VSD(mostly) TGA, HLHS (rarely) are associated. Among the GI anomalies TEF, Duodenal atresia or malrotation, Hirschsprung disease, small bowel atresia are associated. Lumbo sacral dysgenesis, Spinal dysraphism and Currarino's triad (tethered cord, lipomas, syringo Hydro melia) are also associated.

Preoperative investigations were explained in detail as follows:

INVESTIGATIONS

At Birth

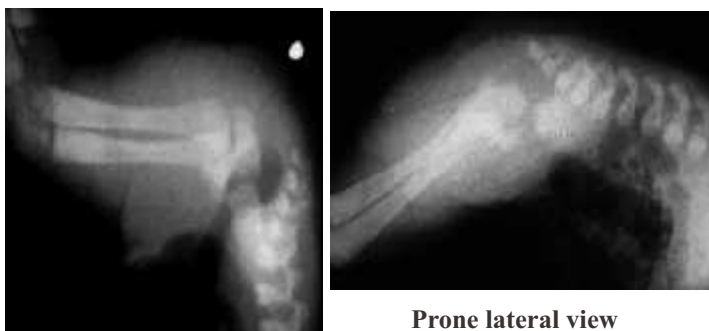
- Invertogram
- X Ray - Prone Lateral view
- X ray Abdomen Erect including Sacrum
- Ultra sound

Before Corrective procedure

- Distal Loopogram / Colograms
- Micturiating Cysto Urethrogram
- Fistulogram, Sinogram
- CT, MRI Not routinely used
- Electro Myography
- Endoscopy

- Wangensteen's Invertogram is done 12 hours after birth (ideally 18 hours in the females & 24 hours in the males) by holding the baby upside down for 3 minutes (usually for one minute in practice). The two trochanters & femur must be super imposed & Xray taken in lateral view with cone of the beam centered on the trochanters. A speck of barium or small metal marker is fixed at the site of anus. Taking X ray image in prone, lateral view of the child is an alternate for Invertogram. The theoretical risk of aspiration & ventricular hemorrhage by inversion of a neonate may be avoided by this. It is more convenient. While taking this X ray, prone position is maintained for some time to allow the gas from the abdomen to enter the rectum.

Wangensteen's Invertogram



Prone lateral view

- X-ray of sacral spine may help to rule out other causes of intestinal obstruction - atresia & meconium ileus and other sacral anomalies X-ray of sacral spine may help predict incontinence due to neurological impairment as mentioned below

Deficiency of 5th and 4th segment - Normal innervations of bladder and rectum with normal muscles.

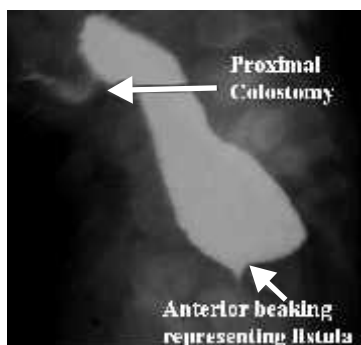
Deficiency of 5th, 4th & 3rd segment - Variable. Most patients are incontinent.

Deficiency of 1st and 2nd segment - Incontinent with poor muscles.

Hemi Vertebra:- Variable.

- Ultrasound is used to rule out renal anomalies & to measure the pouch, perineal distance. The distance is measured as PP length (perineum - pouch length) which denotes the site of the defect - <5mm low, 5-10mm intermediate, > 10mm high. This is not a fool proof test.

- Distal Loopogram / Colograms is done by giving a bowel wash to evacuate faecalomas. Dilute water soluble contrast is used to identify a fistula communicating with the urinary tract. The level of pouch is assessed and the presence of anterior beaking shows the fistula site.



This gives an idea of the distance of the bowel that is to be pulled down. Descending colon colostomy or Proximal sigmoid colostomy are done for high anorectal anomalies.

- Micturating Cysto Urethrogram is performed to identify other urological abnormalities. Fistulas are better seen in colograms.
- Genitogram, Cloacograms are useful in Cloacal anomalies
- CT, MRI are not routinely used. They are used to assess the spinal column & in cases with neurological involvement.
- Electro Myography & Endoscopy-
- Cystoscopy/Vaginocopy are performed as per requirements.

Based on the reports, surgery is performed as per the protocol with regards to low/high lesions respectively.

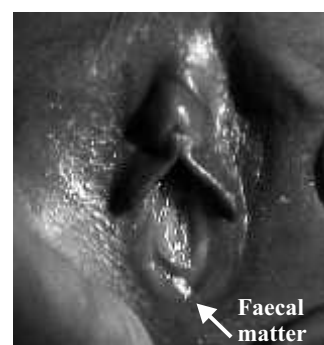
Covered anus complete and incomplete

Posterior Triangular Anoplasty



Ano Vestibular Fistula

Posterior cutback, dilatation and Posterior transposition at 9 months or when the child gains 10 Kg of weight.



Ano Cutaneous Fistula, Anterior Perineal anus, Vulvar Fistula and anus

Dilatation and posterior transposition at 9 months or when the child gains 10 Kg of weight

Anal Stenosis and imperforate membrane

Dilatation

High Anamolies

Colostomy followed by appropriate pull through at 9 months or when the child gains 10Kg of weight predominantly modified Posterior Sagittal Ano Recto Plasty (PSARP) is performed. Pelvic colostomy is preferable. However if the lesion is high & the distal bowel loop would not be sufficient for mobilization, a trans colostomy is performed.



Laposcopic procedures for ARM are still evolving and when standardized would yield good results. He ended saying that all ARM corrected need to be evaluated with specific scoring system in practice. Given below is a scoring card which helps to assess efficacy & the well being of the patient after the surgery.

Kelly clinical "score", is an assessment of the sphincteric action of the "Pubo Rectalis". The maximal score is 6 for a child with normal control under all circumstances, who is always clean and has a strong squeeze of the "Pubo Rectalis" whereas a totally incontinent child can have a score of 0. Point score is as follows (good = 5 - 6 points, fair = 3 - 4 points, poor = 0 - 2 points)

2 points	Normal control under all circumstances, with no accidents
1 point	Occasional escape of feces or flatus
0 points	No control, or frequent accidents (more than 50% of the time)
2 points	Always clean
1 point	Occasionally stained
0 point	Always staines
2 points	Effective, strong squeeze of Pubo Rectalis
1 points	Weak or partial squeeze of Pubo Rectalis
0 points	No contraction detectable

An Appeal



- ☞ Are you an Obstetrician/Neonatologist/Paediatrician/FOGSIAN ?
 - ☞ Have you enrolled in the NATIONAL MOVEMENT called Birth Defects Registry of India (BDRI)?
- If no, please enroll today and contribute your hospital data for a cause.

For details contact

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