Current practice and emerging trends in management of common surgical congenital malformations in neonates

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Abstract
Congenital malformations are significant but underrecognized cause of mortality and morbidity due to disability in neonates in almost every part of the world. Their occurrence, subsequent management and outcome are direct or indirect indicators of health parameters in a community. Current scenario is changing and significant development has been made in early diagnosis and treatment of congenital anomalies. But, this improvement has been largely limited to developed nations. In this review article author shall discuss epidemiology, presentation and current trends in management of common surgically manageable malformations.

Keywords: Congenital malformation, Congenital disorders, Neonates, Birth defects, Fetal medicine, Prenatal intervention.

Introduction
The term congenital malformation or anomaly is often used interchangeably with birth defects or congenital disorder. An update from media center of WHO in 2016, defines congenital anomalies as structural or functional anomalies (for example metabolic disorders) that occur during intrauterine life and can be identified at birth, or sometimes may only be detected later in infancy, such as hearing defects.[1]

Data on incidence of congenital anomalies is sparse, particularly in developing countries. It has been conservatively estimated to be three to six percent.[2] Studies from south India which were limited to tertiary care institutes, earlier reported incidence of 1.97 percent at birth.[3] These figures excluded anomalies appearing later in life and inborn error of metabolisms. Another prospective study from southern India reported overall incidence of 3.7 percent. Still birth deliveries have higher incidence of anomalies. This was reported up to 15.7 percent.[4] This rise in incidence signifies need for autopsy in stillborn babies. The frequencies of occurrence of these anomalies vary from study to study. me study report musculoskeletal anomalies like cleft lip palate, club foot as commonest with 9.69 per 1000 live birth,[4] while others reported central nervous system , neural tube defects as commonest.[5,6] In authors experience as surgeon, neural tube defects, anorectal malformations and musculoskeletal anomalies like club foot, cleft lip and polydactyly are commonest anomaly surgically treated at birth while renal defects like fetal hydronephrosis and cardiac defects like small VSD or PDA are commonly detected antenatally but many do not require surgical treatment.

As evident by some of these studies from southern and central part of India, efforts are now made to formulate a registry like Kerala registry of epilepsy and pregnancy which recently analyzed relative risk of malformations.[7] This attempt may help in assessing exact burden of these malformations in a community. Data collected subsequently may be utilized for formulating policies to manage them.

WHO estimated about seven percent of all neonatal death due to birth defects in 2010.[1] In India these anomalies account for 8-15 percent of perinatal deaths and 13-16 Percent of neonatal deaths.[8,9] Anomalies are reportedly more common in males, preterm babies and those born out of consanguineous marriages. Maternal factors of late age beyond 30-35 years, hypertension, diabetes, smoking habits and teratogenic effects of drugs like valproate and clobazam in embryogenesis phase have been implicated as causes.[7] There are genetic factors and environmental factors involved in etiology of these anomalies.

This article shall deal with current trends in management of some common surgical anomalies in newborn period. This shall include their diagnostics and treatment.

Specific surgical conditions
Anorectal malformations (ARM)
Introduction
ARM are common malformations of gastro intestinal tract managed surgically in neonatal period. These include spectrum of defects in both genders ranging from low anomalies like anal membrane or perineal fistula to more complex anomalies like high common channel cloaca. Earlier many classifications were proposed like wings spread classification, but with passage of time due to confusing terms and new additions, classification was simplified by Alberto Pena.[10] Subsequently it was again modified with involvement of rare and regional anomalies like pouch colon atresia in krickenbeck classification in 2005. (Table I) [11] ARM has traditionally been classified as

high, intermediate and low variety. It is usually associated with communication with urinary tract at different level. Sometimes communication may not exist and then it is termed as ARM without fistula.

Table I: krickenbeck classification for anorectal malformations 2005

Major clinical groups	Rare and regional variants
Perineal (cutaneous) fistula	Pouch colon atresia/ stenosis
Rectourethral fistula Prostatic bulbourethral	Rectal atresia/ stenosis
Rectovesical fistula	Rectovaginal fistula
Vestibular fistula	H-type fistula
Cloaca	others
ARM with no fistula	-
Anal stenosis	-

Epidemiology and etiology

An overall incidence of 1in 2000 live birth to 1 in 5000 live births is reported.[12] but higher incidence has been reported in some southern and eastern part of our country to extent of 1in 600 live births. There is higher incidence of ARM in prematurity, low birth weight and twin pregnancies approximately 25-45 percent cases, this anomaly is isolated defect. About 80 percent of girls and 50 percent of boys have low anomalies.[13] Recto bulbar urethral fistula is the commonest anomaly in boys and recto vestibular fistula is most common among girls. No exact etiology is known for isolated ARMs but genetic predisposition plays some role along with consanguinity in some infants. Those with associated malformations and syndromes have genetic etiology. Recently high incidence of ARM is reported with inutero exposure with lorazepam and reduced incidence with intake of folic acid during pregnancy.[13] But last word is yet to be written about etiology.

Clinical presentation and diagnosis

Ultrasound antenatally is much less sensitive investigation for diagnosing large bowel defects. Since large bowel is mainly a reservoir with no function at this stage of life, defect in this region are difficult to detect. However, presence of echogenic bowel, polyhydramnios, dilated bowel especially in third trimester is suggestive of obstruction at that level.[14]

Mostly ARM in boys and girls is a clinical diagnosis after birth. (Fig. 1, 2) There is absent anal opening at normal place with or without fistulous opening externally. Babies presenting late may have abdominal distention and bilious vomiting and intolerance to feed. In females, especially diagnosis may be delayed or missed in immediate newborn period as girls continue to pass meconium without signs of obstruction. All orifice examination immediately after birth in both genders is therefore a crucial step.

Fig. 1: Female ARM perineal fistula (right) caudal regression syndrome (left)

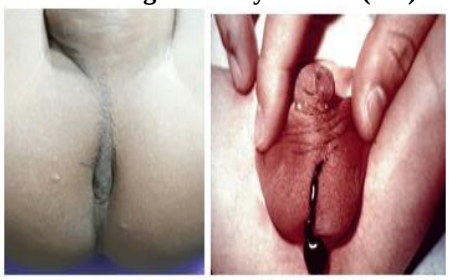

Fig. 2: Male ARM showing anal dimple and absent anal opening (Left) Meconium from fistulous tract (Right)

Post-natal management

Preoperatively, the goals of management are two. A) Assessment of presence of associated congenital malformations in first 24 hours B) Assessment of type of ARM and procedure for correction and timing after 24 hours. Good examination of perineum is key and this provides clue to diagnosis. Presences of meconium over perineum suggest low anomaly and meconium in urine suggest communication at higher level. This may take at least 24 hours to confirm. A perineum with deep natal cleft or gluteal fold with prominent anal dimple is suggestive of intermediate or high with recto bulbar variety in males. While flat perineum is seen in higher anomaly like recto bladder neck fistula in males or cloaca in females.

In female, number of opening in vestibule helps in diagnosis. Three openings in vestibule are commonly seen and third opening is within limit of posterior fourchette. This is vestibular fistula. Two opening with third outside vestibule and outside the limit of sphincter suggests perineal fistula. Very rarely in about 1 Percent or less cases opening may be seen in Vagina.[12] Single opening is seen in cloaca where the common passage or urogenital sinus may be short (less then 3cm) or long (more than 3 cm).

Associated malformations like sacral defects, renal agenesis, vesicoureteric reflux, cardiac defects like ASD, VSD or PDA, other gastrointestinal anomalies like esophageal atresia, duodenal atresia or malrotation may be present and need to be ruled out by following investigations:

- Radiograph of abdomen and chest (baby gram)
- Radiograph of spine or pelvis
- Sonography of abdomen and KUB region

- Echocardiography
- Voiding cystourethrogram (VCUG)

After 24 hours, male with ARM not showing signs of meconium on perineum are subjected to prone cross table lateral radiographs (Fig. 3) to ascertain level of defect based on level of air in relation to aforesaid bony landmarks. Rarely karyotyping may be needed in babies with associated malformations.

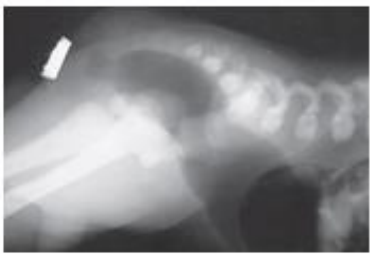

Fig. 3: Prone cross table radiograph showing air contrast just above the marker. Low anomaly

Surgery is mainstay of treatment. These are managed either in single stage or staged procedures depending on type of defect, institutional set up and skill of operating surgeon. The choice of procedure depends on level of defect. In imperforate anal membrane or perineal fistula anoplasty is procedure of choice. In high or intermediate type of anomaly, divided low descending colostomy is recommended.[12] Definitive procedure is deferred till six to eight weeks and done after complete work up of associated anomalies. Before performing these procedures, one important investigation needed is pressure augmented cologram. (Fig. 4)[12] Water soluble dye or thinned out barium is pushed in distal loop of colostomy and still radiographs are taken or fluoroscopy may also be used. This gives clue for level of fistula and avoids injury to surrounding structures like vas deferens during surgery.

In male babies with recto bulbar or recto prostatic fistula, posterior sagittal anorectoplasty (PSARP) is procedure of choice. (Fig. 4) For recto bladder neck fistula, abdominoperineal PSARP is performed. Abdomen is explored to ligate fistula from above.

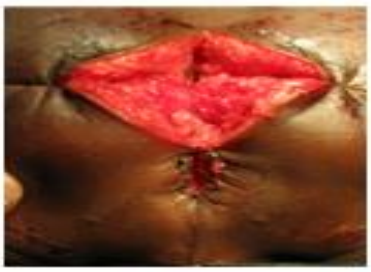

Fig. 4: Operative photograph of PSARP. Anoplasty completed.

The approach is usually by midline sagittal incision by open technique. These are however also done by laparoscopy in which rectum is mobilized till fistulous communication and subsequently excised and tunneled out in midline and anoplasty is performed. However, in personal experience of author, the mobilization above fistula where rectum share common wall with urethra and chances of injury to urethra is high, the dissection is better with open technique and proper repair of fistula is possible without leaving stump. Incision is hidden in natal cleft and hardly visible, cosmesis is also comparable. Therefore, we recommend conventional PSARP to patient. However, in recto bladder neck fistula, communication is 'T' Shaped direct without common wall and therefore proper ligation is possible in laparoscopy.

Presence of sacral anomaly is single important predictor of outcome of bowel and bladder continence. Those with uncomplicated defects have almost 100 percent survival and 75percent have voluntary bowel movements. Urinary incontinence may be seen in females with cloaca repair.

Hirschsprung's disease (HD, congenital intestinal aganglionosis)

Introduction

First described by Harald Hirschsprung in 1888, this condition is genetically determined, surgically correctable lesion in which there is abnormal innervation of gut leading to neonatal intestinal obstruction. Traditionally, HD has been classified as most common type involved recto sigmoid area or short segment HD or classical HD, long segment HD involving area beyond splenic flexure, total colonic aganglionosis, and extended intestinal aganlionosis involving part of distal small intestine. Rarely there may be total intestinal aganglionosis. A condition of ultrashort segment HD is subsequently renamed as internal sphincter achalasia. There may be allied neurocristopathies of gut presenting similarly like intestinal neuronal dysplasia which needs to be histologically differentiated.

Epidemiology and etiology

Overall incidence reported is 1 in 5000 live births but it varies in different ethnic groups. It is most l common surgical cause of lower intestinal obstruction in neonates. Male to female ratio is 4:1 and as length increases the ratio decreases to 1:1 in TCA. Familial cases have more TCA. Enterocolitis is more common in TCA. Great advances have been made in understanding the molecular biology of HD. Mutation in at least nine partially interdependent genes have been detected. These include RET, EDNRB, GDNF, NTN etc.[15] Pathologically there is absence of ganglion cells in myenteric and submucosal plexuses in bowel wall extending up to variable distance from IAS. This happen due to failure of cranio caudal migration of neural crest cells.

Clinical presentation and diagnosis

- HD may present as
- Delayed passage of meconium beyond 24-48 hours of birth
- Chronic Constipation and failure to thrive
- As intestinal obstruction
- As neonatal pneumoperitoneum due to enterocolitis
- Diagnostic armamentarium includes,

Radiograph of abdomen: This may show gas filled distended loops and no rectal gas shadow, or free gas under diaphragm if perforated. (Fig. 5)

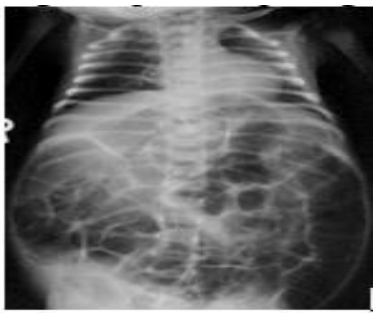

Fig. 5: Abdominal radiograph showing gas filled dilated loops in Hirschsprung's disease.

Barium enema: This is done without prior rectal examination and saline bowel washes. It shows transition zone of proximal dilated colon and gradual narrowing of aganglionic rectum in lateral or oblique film. Reversal of recto sigmoid ratio is also diagnostic of HD. In newborns however, transition zone may not be distinguished clearly, so a delayed film or post evacuation film after 24 hours is considered diagnostic if dye is retained.

Anal manometry: This is done with help of pressure transducer with probes in anal canal. With increase in intrarectal pressure normally there is inhibition of IAS and this is called as recto anal inhibition reflex. Failure of this inhibition reflex suggest HD. But it is less conclusive in infant younger than one month and long-standing constipation.

Rectal biopsy: This is gold standard in diagnosis of HD. It may be full thickness rectal biopsy, done under anesthesia or Suction mucosal biopsy often done as office procedure. It is taken 2cm above dentate line to avoid zone of physiological aganglionosis. Absence of ganglion cells in specimen and presence of hypertrophic nerve bundles in H& E staining is suggestive of HD. Acetylcholine esterase increase activity is seen in muscularis mucosa and decrease or absent calretinin immunoreactive fibers in lamina propria[15-20]

Other test required for associated malformations include

- Echocardiography
- Sonography of abdomen and renal system
- Karyotyping if trisomy 21 is suspected
- Computerized tomography or Magnetic resonance imaging of brain in selected cases.

Treatment

Surgery is the main stay of treatment. It may be done as single stage procedure or staged procedure depending upon type of HD and condition of patient. It may be done as open procedure from abdominal route or trans anal route or with laparoscopic assist. In staged procedure, colostomy with multiple biopsies is performed initially followed by definitive procedure.

Historically three basic procedures were performed in different surgical hands with almost similar long-term results. These include:

- Swenson anorectal pull through
- Duhamel retro rectal pull through
- Soave's endorectal pull through

Presently, single stage trans anal endorectal pull through is widely performed in early age when colonic dilatation is not significant. Need of frozen section biopsy and experienced pathologist is must for performing this procedure. The advantage includes early resumption of feeds, less pain and shorter hospital stay. Long term results however show increase incidence of fecal incontinence as compared to open procedures.[21]

Neural tube defects (NTD)

Introduction

It includes spina bifida occulta and spina bifida aperta. Meningomyelocele (MMC) involves herniation of meninges and neural tissue through vertebral arches openly or in a membrane covered swelling usually in lumber or sacral region. If herniation involves only meninges then it is called meningocele (MC). Approximately 90 percent of open spina bifida is MMC which has damaged nerve tissue and so some kind of neurological deficit and 10 percent is MC with no neurological deficit. Some studies report highest incidence of NTD in Northern Ireland and south wales with incidence of anencephaly is 6.7 per 1000 and spina bifida of 4.1 per 1000.[22] In India however, reported incidence is 0.5 to 11 percent.

Diagnosis

Prenatal diagnosis with sonography at 18-20 weeks and maternal serum alpha fetoprotein (MSAFP) is possible though difficult. MSAFP is elevated in almost 80 Percent of women in mid-trimester carrying fetus with NTD. Post- natal diagnosis is clinical and skin covered or membrane covered swelling is seen in midline over lumbosacral region, cervical, or thoracic region. (Fig. 6) This may rupture with leakage of cerebrospinal fluid. Ultrasound of brain and KUB is done for hydrocephalus and urological abnormalities. Magnetic resonance imaging (MRI) of spine is needed for outlining the detailed anatomy of lesion.

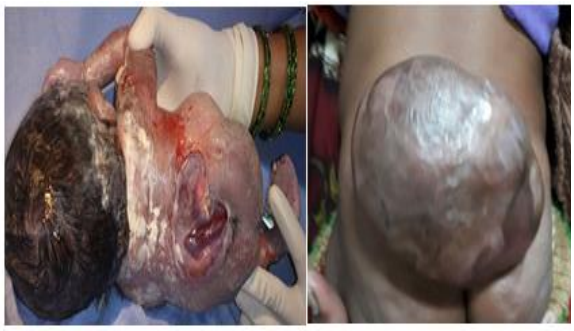

Fig. 6: Clinical photograph showing lumber MMC (Right) and open neural tube defect myelocele (Left)

Treatment

It requires multidisciplinary approach. Many such defect with risk of rupture or leakage may soon be closed immediately after birth by pediatric or neurosurgeon. Hydrocephalus may be present or develop later which require shunt placement or third ventriculostomy by endoscope.

Fetal surgery is also performed by 18-25 weeks for stopping leakage by early excision and repair of MMC. In MOMS trial this showed reduction in need of shunt placement in first year of life as compared to surgery in postnatal period.[23]

Excision and repair of meningocele with intact skin may be done electively, but meningomyelocele with impending rupture or with cerebrospinal fluid leakage needs urgent attention and repair preferably within 72 hours.

Treatment of associated urogenital problems of neurogenic bladder is done by proper monitoring of urinary system. Use of anticholinergics and clean intermittent catheterization is successful in keeping pressure in normal range and avoid higher system failure. Saline bowel enemas or Malone antegrade continence enema (MACE) is done for fecal incontinence or constipation. They9 should be nurtured in latex free environment.

Club foot is managed by physiotherapy initially or use of special shoes or POP or in some case tendon release surgery.

Rehabilitation of such patients is important as there may be lifelong disability. They need proper psychological care from family and peers.

Congenital diaphragmatic hernia (CDH)

Introduction

CDH is a major cause of respiratory failure and therefore high morbidity and mortality in newborns. Its incidence is reported to be 1in 2000 to 1 in 3000 live births.[24] CDH is primarily of four clinical types viz anterolateral hernia due failure of formation of lateral component of septum transversum, posterolateral hernia also known as bochdalek hernia caused by failure of closure of pleuroperitoneal canal, pars sternalis hernia by deficiency in medial retrosternal part of septum, morgagnian hernia by failure of muscle consolidation around foramen of Morgagni. Most commonly, around 96 % cases of CDH are bochdalek hernias. Around 84% are on left side. It may be isolated defect or associated with syndromes CDH may have varying degree of pulmonary hypertension and pulmonary hypoplasia which contribute to high mortality in patients.

Clinical presentation and diagnosis

Antenatally CDH may be diagnosed by sonography at 20 weeks by evidence of bowel in thoracic cavity. Detection rate is approximately 60 percent in developed countries.[24] Fetal lung Head ratio (LHR) less than one and polyhydramnios are considered poor prognostic indicators.

Post-natally baby present with respiratory distress since birth, cyanosis or recurrent respiratory infections and distress lately. Failure to maintain oxygen saturation after birth should alert pediatrician and bag and mask ventilation should be avoided. Orogastric tube should be placed to decompress stomach. Abdomen and chest radiograph will show multi-cystic lesion in lung zones with or without shift of mediastinum. (Fig. 7) There may be absence of rim of diaphragm and bowel air continuity may be seen travelling to thorax. On right side lesions, hepatothorax may be seen. Putting water soluble dye through orogastic tube may also help in diagnosis in difficult case.

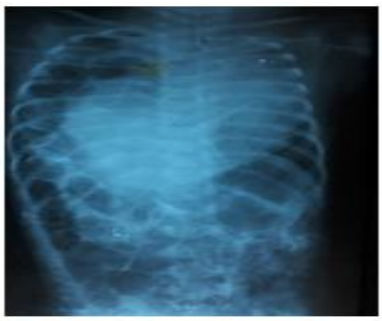

Fig. 7: Radiograph showing absent right rim of diaphragm and bowel herniation in right chest.

Treatment

CDH in current treatment methodology is not a surgical emergency. However, it is a physiological emergency which requires urgent measures to oxygenate post-ductal tissues and reversal of vicious circle of hypoxia and vasoconstriction. Baby should be gently ventilated with low pressures settings to avoid barotrauma of noncompliant lungs. There is spectrum of good patients and patients with poor prognosis.

Delayed surgery is a rule in present times and indications for surgery in ventilated patients include arterial blood gas report with PH in range of 7.35-7.45, arterial PaO2 of 100% at least once in a day and PaCo2 of less than or equal to 40%. This may not be possible always due to hypoplasia and slow correction of

pulmonary hypertension. Other measures used are inhaled nitric oxide therapy, surfactant therapy, sildenafil, HFO ventilation with varying degree of success.

Surgical procedure may be done by open exploration by laparotomy and repair of diaphragm after reduction of all contents gently, or by thoracoscopically in relatively older patient who has stable preoperative parameters. Repair is done by non-absorbable suture or mesh if defect is large.

Fetal surgery for CDH is not encouraging in last two decades. Randomized studies are needed for fetal tracheal occlusion (PLUG-plug the lung until it grows) and EXIT (Ex utero intrapartum treatment). ECMO in Cochrane study in 2002 have not shown additional befits in CDH.[24] At present best fetal surgical treatment appears to be fetendo clip or balloon occlusion of trachea by fetal endoscopy with better survival rates.[24]

Esophageal atresia with tracheoesophageal fistula

Introduction

This foregut anomaly characterized by abnormal communication between trachea and esophagus, often test the skill of individual surgeon and adequate experience is needed to handle these patients. Approximate incidence of this condition is 1 in 3000 live births. This has been classified in to distinct types. There is anatomical classification by gross and prognostic classifications by spitz. (Table 2) Type C defect is most common and is seen in 85-90% cases. Incidence of associated malformation is quite high and range from 50 to 75percent.[25] Type A have highest incidence. Most commonly seen are cardiac anomalies followed by GIT anomalies. These may be summed up in VACTERL and CHARGE syndrome. Associated anomalies play significant role in prognosis of condition.

Table II: Okamoto modification of Spitz risk classification of Esophageal atresia and tracheoesophageal fistula

Class	Description	Risk	Survival %
I	No major cardiac anomaly BW > 2 kg	low	100
II	No major cardiac anomaly BW < 2 kg	moderate	81
III	major cardiac anomaly BW >/ = 2 kg	Relatively high	72
IV	major cardiac anomaly BW < 2 kg	High	27

Clinical presentation and Diagnosis

Prenatally this condition is rarely suspected and mostly diagnosed postnatally. However, presence of polyhydramnios and absent stomach bubble along with dilated esophageal pouch should alert clinician. These two have positive predictive value of 56 percent.[24] Postnatally, failure to insert orogastric tube beyond 10cm is important sign. There is respiratory distress and excessive frothing of saliva as baby is not able to swallow it. Chest radiograph taken with soft red rubber catheter in place shows limit of blind ending upper pouch in lateral view. (Fig. 8) Isolated atresia shows gasless abdomen. (Fig. 9) Echocardiography is needed for heart defects and right sided aortic arch. Sonography of abdomen for GI anomaly and urogenital anomalies.

Fig. 8: Lateral radiograph showing soft red rubber tube hold up at T4 T5 junction

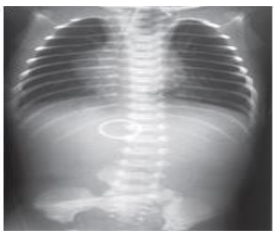

Fig. 9: Abdominal radiograph showing gasless abdomen in isolated esophageal atresia.

Treatment and prognosis

Preoperative stabilization of these babies is important. This includes neonatal intensive care (NICU), frequent low-pressure suction of upper pouch, oxygenation, elective ventilation if needed and prematurity management. Babies with adequate birth weight with 2.5 kg with no pulmonary complications and no serious cardiac defects can undergo surgery for primary repair. The procedure is right sided posterolateral thoracotomy through fourth intercostal space preferably extra pleural approach and ligation of fistula with end to end esophageal anastomosis. (Fig. 10). Post-operative dye study is done to confirm no leak before starting feeds. (Fig. 11)

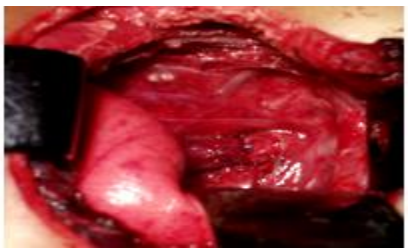

Fig. 10: Operative photograph of thoracotomy showing esophageal anastomosis over infant feeding tube.

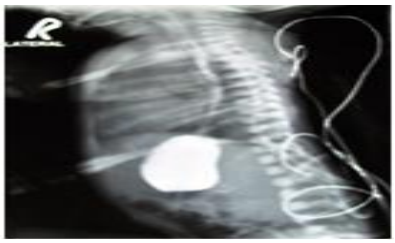

Fig. 11: Post- operative dye study after successful esophageal anastomosis.

Others with serious defects or low birth weight of 1.5kg or below or with pulmonary complication are subjected to parenteral nutrition, upper pouch suction, gastrostomy until they are fit for procedure.

This procedure in neonates now a day may also be done by 2mm thoracoscopy instruments, but case selection is important.

Babies with isolated atresia are managed with esophagostomy and gastrostomy initially, followed by esophageal reconstruction. This may be done as primary procedure at birth or later in life. Variety of organs may be used for esophageal replacement. None of them however is ideal. Gastric transposition, gastric tubes from greater curvature or colonic transposition are done with comparable long-term results. These procedures are also done by laparoscopy assisted technique.

Complications include leakage, stricture, reflux, recurrent fistula and tracheomalacia. Survival rate may be more than 90 percent in babies with good weight and no associated anomalies, while it may be 25 to 65 percent in other category with low birth weight and associated anomalies.[25]

Upper urinary tract obstructions

Introduction

Hydronephrosis or fetal urinary tract dilatation is commonly detected renal anomaly in antenatal ultrasonography. It accounts for 0.6-0.65 percent of pregnancies.[24] Not all dilatations, however suggest obstruction to fetal urinary tract. Most of these dilatations are transient or physiological. These resolves as gestational age progress. However, they require postnatal monitoring. Most common pathological cause of upper urinary tract dilatation is pelviureteric junction obstruction (PUJO).

Epidemiology and Clinical presentation

Reported incidence of PUJO is 1 in 500 live births. Boys are affected more than girls, left side kidney is more frequently involved and approximately 10 percent cases are bilateral.[26,27] Mostly with registered cases, antenatal sonography detects hydronephrosis and presentation is asymptomatic. Different grades of hydronephrosis are assigned depending upon degree of dilatation. Society of fetal urology (SFU) grading is commonly used. (Table III) Also, measurement of transverse pelvic diameter (TPD) along with lower pole parenchymal thickness and their ratio is more specific for screening of these babies and plays vital role in decision of surgery. TPD of up to 10 mm is considered mild hydronephrosis, 10- 15 mm is moderate and above 15- 20 mm it is considered severe hydro nephrosis in infants. (Fig. 12)

Table III: Society of fetal urology grading for antenatal hydronephrosis

Grade 0	No dilatation, calyceal wall appose to each other
Grade 1 mild	Dilatation of renal pelvis without dilatation of calyces. No parenchymal atrophy
Grade 2 mild	Mild dilatation of pelvis and calyces No parenchymal atrophy
Grade 3 moderate	Moderate dilatation of pelvis and calyces Blunting of fornices, mild cortical thinning
Grade 4 severe	Gross dilatations of renal pelvis and calyces, ballooning Loss of borders between renal pelvis and calyces Renal atrophy seen as cortical thinning

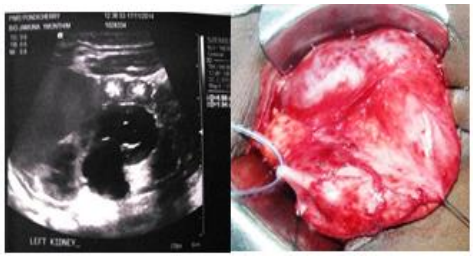

Fig. 12: Sonography of left kidney showing dilated renal pelvis and calyces(Left).Operative photograph of pyeloplasty showing dilated pelvis and PUJO.

Although uncommon these days, but child may present with smooth abdominal mass, urinary tract infections or in severe cases with urosepsis. These presentations are still seen in hospitals catering rural or semi-urban India and author has experienced it in an area where population is not aware for need of antenatal sonography.

Treatment

Those patients requiring surgical treatment are usually having moderate to severe hydronephrosis with TPD more than 20 mm and renal scan showing fall in glomerular function rate and renal function below 40 percent. Renal scan also shows obstructive wash out curve in graphical form with increased T½ half-life of radiotracer beyond 15-20 min. Other associated anomalies like multicystic dysplastic kidney, vesicoureteric reflux or renal agenesis (solitary kidney) are to be investigated preoperatively.

Procedure of choice for these patients is Anderson hyne's dismembered pyeloplasty. (Fig. 12) This may be done by open technique by different approaches or by laparoscopic assisted pyeloplasty in infants. In older children, it may be performed as Tran's peritoneal

pyeloplasty or retroperitoneal pyeloplasty. Basic principle remains same in all forms as adynamic segment of ureter is excised and mucosa to mucosa anastomosis of ureter to pelvis is done in same axis. Postoperative follow up is needed with renal scan to assess improvement or stability of functions.

At centers with availability recently, robotic pyeloplasty is also gaining popularity. Experience of author in children with other alternative technique like percutaneous and retrograde endo-pyelotomy is limited.

Lower urinary tract obstructions

Introduction

Posterior urethral valves (PUV) or congenital obstructive posterior urethral membrane (COPUM) as it is called is most common cause of lower urinary tract obstruction and bilateral obstructive uropathy in male newborns and infants. They have high morbidity and mortality due to associated renal dysplasia and pulmonary hypoplasia. Those with poor renal functions develop end stage renal disease and require renal transplant. In 1919 young described three types of PUV based on cystoscopic appearance. Types I valve which originate distal to verumontenum and diverge anterolaterally are the most common variety. Type II is considered artifact and is of historical importance now, whereas type III is centrally perforated diaphragm and is responsible for five percent of obstruction.

Epidemiology and clinical presentation

Incidence is variable reported in range of 1 in 5000 to 1in 8000 live birth and highest incidence is reported from Oman of more than 1in 250 births. Consanguinity is noted in majority of cases.[28-30] Most of the patients are diagnosed antenatally or in newborn period is suspected in fetus with bilateral hydronephrosis and thick-walled bladder and posterior urethra. 'Key hole' sign has been concluded as less specific in some studies.[31] There may be oligohydramnios.

In areas where antenatal screening program are not intrinsic part of working system, detection may be late and baby may present with poor stream or dribbling of urine, failure to thrive, Urosepsis, recurrent urinary tract infections, urinary ascites, and fever. PUV has spectrum of cases, prognosis depends on associated dysplasia, renal functions and pulmonary complications.

Treatment

PUV patient is initially catheterized with soft indwelling catheter like infant feeding tube and bladder is decompressed. Urine analysis is done for infection and microalbuminuria. Fluid and electrolyte correction is done and intravenous antibiotics are started. Renal function test particularly serum creatinine is done which may be high initially.

After initial catheterization, and hydration of baby, voiding cystourethrogram is done to asses' vesicouretric reflex, its laterality and grade. (Fig. 13) This investigation also confirms presence of dilated posterior urethra with abrupt cut off suggestive of valve. Cystoscopy with valve fulguration is done by hook electrode or cold knife cutting. (Fig. 13) In high grade reflux and severe dilated tortuous ureter, upper system may need urinary diversion like vesicostomy, ureterostomy or pyelostomy. Valve fulgurations are also done by laser. Use of mechanical valvotome a device to cut the valve is option available if facility for fulguration or expertise is not available. However, this is blind procedure and risk of injury to newborn urethra is there in inexperienced hands.

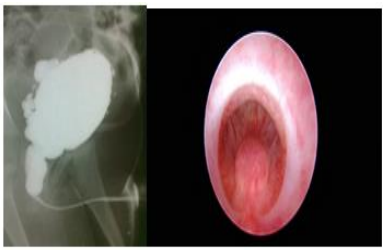

Fig. 13: VCUG showing dilated posterior urethra and bladder diverticula and abrupt cut off (Left), Cystoscopic view of type I valve, distal to verumontenum. (Right)

After valve fulgurations babies are monitored for residual urine and since bladder in non-compliant, need of anticholinergic medicines and clean intermittent catheterization (CIC) are used to reduce bladder pressures. Use of alpha blockers like terazosin is helpful in reducing post void residue due to its action on bladder neck, it improves urinary stream. Those patient with noncompliance for CIC, therefore benefit from these drugs. These procedures have reduced early mortality in newborns from 50 percent to 1-3 percent.[30]

Fetal procedures of vesico-amniotic shunting or fetal vesicostomy is met with variable degree of success. Those fetuses diagnosed before 24 weeks carry poor prognosis and these procedure does not improve their renal function postnatally.[32] More multicenter trial studies are therefore needed to pinpoint group of patients and timing for interventions for maximum benefit.

Sacro coccygeal teratoma(SCT)

Introduction

Teratomas are embryonic tumors containing tissues from all three germ layers derived from totipotent cells. They arise from germ cells or somatic cells. Teratomas are usually seen in midline or paramedian plane from brain to Sacro coccygeal region. SCT is the most common neonatal tumor with substantial risk of obstructed labor. Its incidence is 1 in 35000 to 1in 40000 live births.[24] Mostly at birth these may be benign lesions but as the age progresses tumor may become malignant or undifferentiated.

Classification and presentation

SCT is usually classified in four types (Altman classification).Presentation and prognosis vary with type of tumor. Type 1 2 and 3 can be diagnosed in new born period and need to be differentially diagnosed with other skin covered defects at birth like meningocele or lipomeningomyelocele. Type 4 is usually detected late when child present with recent onset of constipation and digital rectal examination reveals a presacral mass. This is often seen in girls.

Diagnosis

Doppler sonography is important diagnostic tools. MRI postnatally or in fetus can reveal intrapelvic component. SCT can be diagnosed in antenatal sonography and those diagnosed before 30 weeks and associated with polyhydramnios carry poor prognosis.[24]

Treatment

Main stay of treatment for type 1 and 2 is surgical excision at birth. Neoadjuvant or adjuvant chemotherapy is required in large tumors with malignant degenerations before or after surgery. Chemotherapy drugs used are vincristine, methotrexate, and actinomycin-D. These may also be given in instance of incomplete excision. Approximately 80 percent of lesions are benign at birth. Malignant degeneration is seen commonly in older children beyond three months. Tumor become hard and skin overlying may give away and ulcerated with discharge of tumor tissue. This carry poor prognosis.

At birth, large vascular tumors may have situation of high out cardiac failure and anemia in newborns or also in fetus leading to fetal hydrops. Rupture of tumor may cause significant bleeding and mortality of almost 100 percent.[24]

Surgery is done by sacral incision however in some cases abdomino-sacral approach is needed. Removal of coccyx and control of median sacral vessels is mandatory step. Preoperative and post-operative evaluation and screening of tumor and its recurrence is done by serum Alpha fetoprotein (AFP). Its half-life is 6 days and postoperative fall in AFP is a good sign. Persistent elevation for four to six weeks following surgery or rise after initial fall, mandates imaging study to assess for recurrence.

For recurrence etoposide and cisplatin are used along with excision of residual or recurrent tumor tissue. There may be metastasis to brain, lung and liver from adenocarcinoma from epithelial cells.

Fetal surgery is also done for SCT. This involves tumor resection or ablation of feeding vessel particularly in hydrops patient. However antenatal diagnosis and surgery in newborn period with complete excision usually have good outcome as benign tumors do not require chemotherapy. This require assessment studies for need of fetal intervention in SCT and probably only indication is fetal hydrops.

Role of pediatric surgeon and current scenario in India

The role of pediatric surgeon in management of congenital malformations is multipronged. To council parents about natural progression and outcome of anomaly, surgically treat it either by prenatal interventions or postnatally depending on disease, facility and training available. Pediatric surgeon must also co-ordinate and collaborate with other departments like obstetrician, geneticist, neonatologist, social coordinator, radiologist and pathologist.

In developing countries like ours, fetal medicine is emerging as new entity now. In India, some centers in south India at Kochi and Chennai have started prenatal interventions. Risk to mother, preterm abortions and economic issues are limiting factors for its success in India. Main stay in management of many such anomalies therefore, remains surgery in newborn or infantile period. So many anomalies which were fatal earlier are now surgically treated in newborn period with almost 90% success rate. These surgeries may be performed as single stage procedures or staged correction depending on set up, skill of surgeon and patient selection criteria

The horizons of pediatric surgical practices have been expanded due to prenatal surgical diagnosis. Presently anomalies like twin- twin transfusion syndrome due to anomalous vessels are satisfactorily treated antenatally by laser. While others like fetal vesico-amniotic shunt or fetal vesicostomy in posterior urethral valves, PLUG and EXIT in congenital diaphragmatic hernia, excision of neural tube defects and others have met with variable success rate at selected centers in western world. The advantage of antenatal detection by sonography have revealed hidden mortalities in some anomalies like CDH, cystic lymphangioma, or Sacro coccygeal teratoma and timely referral to a center with available facility. The other side of coin is that, this often creates lot of anxiety to parents. So, counselling therefore should be meticulous, ethical and evidence based.

Conclusion

To conclude, though the economic and psychosocial burden due to birth defects is extensive worldwide, birth defect monitoring programs in western literatures has shown downward trend in occurrence of these defects due to early diagnosis by prenatal sonography and prenatal interventions with option of termination of pregnancy as practiced in neural tube defects.

Current scenario in India is also changing and many anomalies which were fatal earlier can now be surgically corrected with gratifying results. But we need proper execution of primary prevention methods like fortification of diet of women bearing age with folic acid and vaccination. We also need standard operating protocols and guidelines for management of these

anomalies at every center involved in tertiary care of newborns. Economic issues however crop up in management of these anomalies and policy formulation by government about inclusion of babies with congenital anomalies in insurance coverage is also urgently needed in our country.

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