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CASE SERIES: A REVIEW OF SOME RARE MANIFESTATIONS AND ASSOCIATIONS OF ANORECTAL MALFORMATIONS.

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ABSTRACT **Introduction:** Anorectal malformation (ARM) is a spectrum disease which affect the distal rectum and anus and also the urinary and/or genital systems with or without associated anomalies. Types of ARM decide the management and surgical repair. We review several types of anorectal malformation with rare and unique presentation. **Case presentation:** We review 7 cases of anorectal malformations with varied presentations operated at TRIHMS, Naharlagun, Arunachal Pradesh. **Discussion:** Anorectal malformation is quite common and many times they are associated with other congenital malformations. clinicians need to examine the ARM babies very closely to see for fistulas, level and type of malformation and also for associated malformations. **Conclusion:** Based on our experience, we strongly recommend a proper evaluation of any anorectal malformation patient for presence of other associated malformations.

KEYWORDS : Anorectal malformations, Anorectoplasty, Colostomy, Congenital pouch colon, Associated anomalies and rectoperineal fistula.

INTRODUCTION:

Anorectal malformations (ARMs) also known as imperforate anus are birth defects in which the anus and or rectum is malformed or absent. ARMs are a spectrum of different congenital anomalies ranging from minor lesions to complex anomalies. They may be associated with other anomalies. Exact etiology of ARMs is not known. Diagnosis is by clinical examination. Nowadays, prenatal diagnosis is possible by expert sonologist. Depending on the degree or level of rectal pouch, it can be classified as low, intermediate and high variety. The higher the level, more complex the treatment. Treatment of anorectal malformations are simple anal dilatation, anopasty, anorectoplasty, anorectourethroplasty, anorectovaginourethroplasty and or treatment of associated anomalies. Surgery may be one time or a staged procedure in which colostomy is performed before definitive surgery and colostomy closure as the last procedure.

Congenital pouch colon (CPC) in which the lower portion of colon end as a blind pouch , is a rare variant of anorectal malformations (ARM) with report of higher incidence in India. Anorectal malformations (ARM) occur in approximately 1 in every 5000 live births while intestinal atresia (IA) incidence is estimated to range from 1.3 and 3.5 per 10,000 live births. The simultaneous presence of both anomalies in one patient is particularly rare. The simultaneous occurrence of Hirschsprung disease (HD) and anorectal malformation (ARM) is extremely rare, with only a very limited number of cases published in the literature. Goldenhar's syndrome is a rare condition with combination of anomalies: dermal epibulbar cysts, auricular appendices and malformations of the ears and vertebral anomalies-oculo-auriculo-vertebral (OAV).

The urogenital sinus is an anomaly with a common channel for the urethra and vagina whereas cloaca is a single opening for the urethra, vagina and rectum. These are extreme form of anorectal malformation.

PRESENTATION OF CASES:

CASE-1. 2 days male new born with Anorectal malformations and rectal atresia. Resection of congenital pouch colon and excision of rectal congenital pouch colon type 4 was done.

Laparotomy of membrane and primary pull through was carried out (Fig.1 & 2).

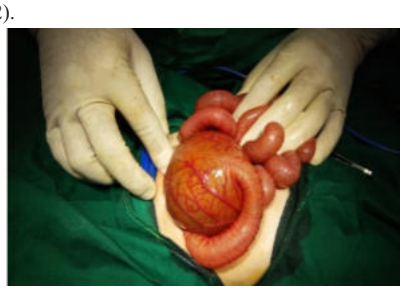


Fig.1: Congenital pouch colon

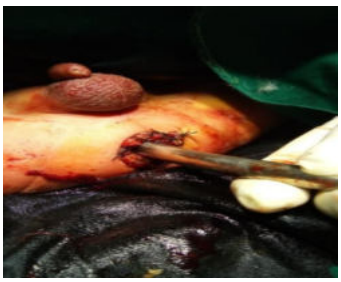
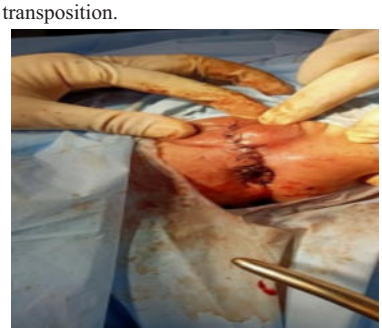


Fig.2: Primary pull through, Anorectoplasty

CASE-2: ARM WITH PERINEAL/ SCROTAL FISTULA.

A newborn male baby with Anorectal malformations and perineal/scrotal fistula. Baby was passing stool through the scrotum. Anal transposition (anorectoplasty) was done (Fig.3).

Fig.3: Anal transposition.



CASE-3: Anorectal malformation with Goldenhar syndrome (oculo-auriculo-vertebral defect) (Fig.4). Anorectoplasty was done (Fig.5).

Fig.4

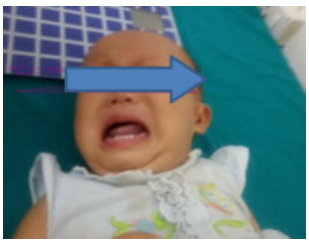
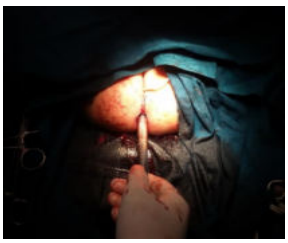


Fig.5

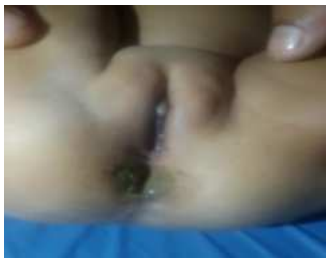


CASE 4: ANORECTAL MALFORMATIONS WITH AGANGLIONOSIS (HIRSCHSPRUNG DISEASE).

Baby presented with Anovestibular fistula in newborn period and anorectoplasty was done. Patient presented at later age with

complaints of constipation. On examination, child had distension of lower abdomen. On per rectal examination- anal stenosis was found, admits little finger only and blast sign was positive. Rectal wash given with anal dilatation done and Glycerine solution instilled. Rectal biopsy showed absence of ganglion cells. Swenson's abdominoperineal pull through operation was done (Fig.6).

Fig.6: Anovestibular fistula.



Case 5. ARM with absent Anal membrane.

A three days old male baby presented with absent anal opening, not passed stool, distension of abdomen & vomiting. Anal dilatation was done on 3rd day (Fig.7, Fig.8, Fig.9).

Fig.7.

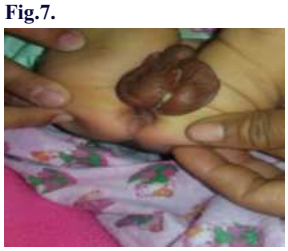


Fig.8.

Fig.9.

Case 6: Cloaca, haemangioma abdomen and generalised congenital abnormal pigmentation.A 3 days female baby presented with single perineal opening. Right transverse colostomy was done (Fig.10 & Fig.11).

Fig. 10.

Fig. 11.

CASE 7: ARM WITH DUODENAL ATRESIA WITH VENTRICULOSEPTAL DEFECT.

1 day old male baby with low ARM with duodenal atresia with VSD. Anorectoplasty , laparotomy and duodenojejunostomy done.

Fig.12: Low ARM.

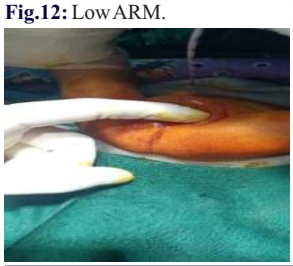


Fig.13: Duodenal Atresia.

DISCUSSION.

Anorectal malformation [arm] is common congenital disease which is prevalent worldwide. it can occur independently or in association with other congenital malformations. Unless we examine the babies properly, anorectal malformation or the associated malformation may be missed.

Many authors have published different types of rare manisfestations and rare association of ARM.

Emily j Gunderson et al reported rare case of anorectal malformation and intestinal atresia.

Congenital pouch colon is rare variant of ARM but relatively more incidence is reported in india. Francesco fascetti-leon et al reported 5 cases of resection of congenital pouch colon.

Raphael N Vuille-Dit-Bille et al mentioned that the association of Hirschsprung disease and anorectal malformations is uncommon association. In our institute, we have come across one such case.

Soichi Shibuya reported Atypical recto-scrotal and recto-perineal fistula in male anorectal malformations: we have come across some cases of perineal fistula, rectoscrotal fistula and rectoperineal fistula. Another rare condition is Goldenhar syndrome. C Seethalakshmi Ashokan reported 2 patients of Goldenhar syndrome in their case series. M Voisin et al published 21 cases of Intestinal malformations and congenital heart diseases in their series. In our institute, we have also encountered good number of cases of intestinal atresia with congenital heart diseases .

CONCLUSION.

Based on our experience, we strongly recommend a proper evaluation of any anorectal malformation patients for presence of other associated malformations like congenital heart diseases, intestinal atresia, musculoskeletal defects, orofacial defects or vacterl association. Thorough clinical examination and proper investigations to ascertain the type and level of malformation and also to rule out associated malformations is very important. Timely treatment is very crucial for the survival of baby in form of medical and surgical management. In this series , all the cases were operated. No death occurred in our series.

CONFLICT OF INTEREST: No conflict of interests

AUTHORS CONTRIBUTION: The authors contributed from the beginning to the end.

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