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Transperineal Ultrasonography in Infants with Anorectal Malformation

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Abstract: ***Background and Objective: Anoractal malformation is a complex spectrum of anomalies. There are 3 tyes of anoractal malformation high, intermediate and low. The distinction can usually be made on the basis of clinical data regarding the presence or absence of a visible perineal opening or passage of meconium through the vagina or urethra This study was conducted to assess usefulness of Transperineal ultrasonography (TPU) as an a non-invasive imaging technique in detection of the type of anomaly, presence and location of fistula. Results: The findings of transperineal ultrasonography (TPU) were correlated with gold standard surgery (Perineal anoplasty/PSARP).The sensitivity, specificity, PPV, NPV and diagnostic accuracy of TPU was 87.5%, 100%, 100%, 77.77% and 91.307% respectively in detection of fistula in ARM. Conclusion: Anorectal malformations are complex spectrum of anomalies predominantly seen in male infants (82.35%) with male to female ratio of 4.6:1.Transperineal ultrasonography (TPU) is a valid and an accurate non-invasive imaging technique in detection of the type of anomaly, presence and location of fistula.***

Keywords: anoractal malformation, transperineal ultrasonography, perineal anoplasty

1. Introduction

Anorectal Malformation

- Complex spectrum of anomalies with Incidence of approximately **1 in 5000 live births.**
- More common in the male infants.[1,2]
- **High type:** hindgut ends above the level of the puborectalis sling.
- **Low type:** passes below the puborectalis sling.
- **Intermediate type**: terminates just at the level of puborectalis sling.
- The distinction can usually be made on the basis of clinical data regarding the presence or absence of a visible perineal opening or passage of meconium through the vagina or urethra.[3]
- Presence of anocutaneous fistula suggest low type of ARM. Absence of anocutaneous fistula/ passage of meconium through the urethra or vagina suggest high/intermediate type of ARM.

Management

- Optimal surgical management - depends on accurate determination of the level of the rectal pouch.
- High/intermediate type- Diverting Colostomy f/b Posterior sagittal anorectoplasty (PSARP).
- Low type- Perineal anoplasty/dilatation of ectopic anus

2. Material and Methods

Study Period: August 2016 to December 2016.

Study Population: 34 infants (28 males, 6 females; median age, 1.78 days; range, 0-4 days) with the diagnosis of anorectal malformation were examined with transperineal ultrasonography to determine the type of anomaly. Two cases were excluded from the final results of the study (n=32), as these patients were died and surgical confirmation could not be done.

The final diagnosis, including the type of anomaly was made on the basis of imaging and surgical findings according to the international classification of anorectal anomalies, which is based on the relationship between the level of distal rectal pouch and the pubo-rectalis sling of levator ani muscle.[4]

Imaging

Prone Cross Table Lateral Radiograph: done 12–24 hrs after birth (to allow gas to reach the distal rectum). Baby was placed in the prone position (genu-pectoral position) for 3 min before taking the radiograph to allow gas to displace meconium and rise to the termination of the pouch.

Transperineal Ultrasonography (TPU): was performed using High-resolution 12 MHz linear array transducer with transperineal approach as described by Teele and Share.[5]

- **Position:** supine position without specific preparation.
- **Scanning:** Mid-sagittal plane through the perineum - used to identify the base of the bladder, the urethra and the vagina/corpus spongiosum (CS) between the urethra and distal rectal pouch. (Fig 1)

Figure 1: Midline sagittal sonogram in normal 3 week old boy, Photograph shows placement of linear transducer.

Distal Rectal Pouch was identified by presence of hypo-echoic meconium or gas within the distal rectal pouch. Scanning was performed in resting state while the child was not crying.

Care was taken not to press/indent the skin (diminish the distance between the distal rectal pouch and the perineum. The distance between the distal rectal pouch and the perineum (P – P distance) was measured in millimeters. Presence of any fistulous communication (suspected by presence of meconium/gas in the urethra or in vagina) between the distal rectal pouch and urogenital system was also noted.

Thereafter, the P – P distance and any associated internal fistula observed on TPU were compared with the type of anorectal malformation confirmed on surgery (Perineal anoplasty/PSARP).

Transperineal Ultrasonography With Infracoccygeal Approach was done to know the thickness and the relationship of puborectalis muscle with the distal rectal pouch. (Fig 2)

Figure 2: Transverse infracoccygeal sonogram in normal 3 week old boy, Photograph shows placement of linear transducer between the anus and coccyx

The puborectalis muscle thickness was measured in millimeters and recorded in medical report.

Transabdominal Ultrasonography was done in all infants using 3-6 MHZ convex transducer to look for any urogenital anomalies.

Distal Colostogram was done before final repair to determine the exact level of anomaly and associated internal fistulous communication

Micturating Cystourethrogram (MCU) was done in selective cases to evaluate associated vesico-ureteric reflux (VUR).

3. Results

The findings of transperineal ultrasonography (TPU) were correlated with *gold standard surgery (Perineal anoplasty/ PSARP).* Thus data was analyzed by using SPSS version 20.0 (trial version).

Appropriate statically test was applied (Kappa for agreement between TPU and surgery).

Sensitivity, specificity, PPV, NPV and diagnostic accuracy of TPU in detection of fistula was also measured by using SPSS soft ware.

On the basis of transperineal ultrasonography (P – P distance), low type of anomaly was detected in 28.12%, intermediate type in 37.5% and high type anomaly in 34.37% infants.

Table 1: Comparision of P-P distance with type of Anomaly (n=32)

Type of anomaly	No. of cases	Mean P-P distance (in mm)	Range (in mm)
Low	9	3.95±1.49	2-7
Intermediate	10	12.7±1.5	10.4-14.2
High	13	20.4±4.7	15-30

- TPU correctly predicted the level of distal rectal pouch in 28 of 32 patients.
- Measure of agreement (kappa) between TPU and surgery (perineal anoplasty/PSARP) was calculated to be 0.812 (p= 0.001).

Table 2: Comparision of TPU and surgery (Perineal Anoplasty/ PSARP) for type of anomaly(n=32)

Type of anomaly	No. of cases on TPU	Actual no. of cases on surgery	No. of cases correctly detected on TPU (surgical confirmation)
Low	9	9	9
Intermediate	12	10	9
high	11	13	10

- TPU detected the fistulous communication between the distal rectal pouch and urogenital system.
- M/c fistula in male - Recto-urethral fistula (66.66%, 8/23)
- M/c fistula in female - Recto-vestibular fistula (4/4).

Table 3: Association of fistula in intermediate/high type of anomaly (n=24)

Type of anomaly	*No. of cases*	*Fistula in no. of cases*	*Percentage*
Intermediate	10	4	40%
high	13	12	92.30%

- Low type of ARM showed Well developed puborectalis muscle and in high type of ARM, puborectalis muscle was poorly developed.

Table 4: Comparision of type of anomaly with puborectalis muscle thickness

Type of anomaly	No. of cases	Muboractalis muscle thickness(in mm)
Low	9	1.4±0.1
Intermediate	10	0.83±0.3
High	13	0.5±0.1

4. Discussion

ARM – incidence of 1 case per 5,000 neonates.[1]

All surgically proven cases of low ARM had P-P distance of $\leq$*7 mm* and transperineal ultrasonography correctly detected all cases of low ARM.

The infants with intermediate type of anomaly had P-P distance of $\geq$ *10.4 mm* and high type of anomaly had $\geq$ *14 mm*

A considerable overlap between the intermediate and high type of anomaly was seen.

There were no false negative results in the present series.

Transperineal sonographic findings (P-P distance) may not be accurate in infants in whom the distal rectal pouch is decompressed by a large fistula.

It is also important that child is not crying while performing the sonogaphic examination (reducing the distance between the distal rectal pouch and the perineum).

In present series, out of 23 cases of intermediate and high type anomaly, 16 infants had fistulous communication with urogenital tract and 14 infants were correctly diagnosed on TPU. In 2 patients with recto-urethral fistula (confirmed on distal colosto-gram and surgery), the fistula was not detected on TPU.

The puborectalis muscle was well developed in low type and Poorly developed in high type of ARM.

In present series the associated anomalies (vertebral, tracheo-oesophageal, musculo-skeletal and urological) were detected in 32.35% (11/34) patients.

The limitation of our study included small sample size, institutional case selection bias and intra-observer and inter-obsever variability of measurements. Further work with series of infants with anorectal malformation is necessary to overcome these shortcomings

5. Conclusion

Anorectal malformations are complex spectrum of anomalies predominantly seen in male infants (82.35%) with male to female ratio of 4.6:1.

Transperineal ultrasonography (TPU) is a valid and an accurate non-invasive imaging technique in detection of the type of anomaly, presence and location of fistula.

The sensitivity, specificity, PPV, NPV and diagnostic accuracy of TPU was 87.5%, 100%, 100%, 77.77% and 91.307% respectively in detection of fistula in ARM.

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