ORIGINAL RESEARCH PAPER

Volume-7 | Issue-5 | May-2018 | PRINT ISSN No 2277 - 8179

INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

A STUDY ON CONGENITAL MALFORMATIONS OF GENITOURINARY SYSTEM IN PERINATES

Anatomy

Hari Charan Sarangsa — Assistant Professor, Department of Anatomy, Silchar Medical College, Silchar, Assam, India.

Turlika Sinha* — Associate Professor, Department of Anatomy, Silchar Medical College, Silchar, Assam, India. *Corresponding Author

Bijon Chandra Dutta — Professor and HOD, Department of Anatomy, Silchar Medical College, Silchar, Assam, India.

Krishna Kanta Biswas — Demonstrator, Department of Anatomy, Silchar Medical College, Silchar, Assam, India.

ABSTRACT

Background: Congenital anomalies of the genitourinary system are found in approximately 10% of infants. Congenital anomalies of kidney and urinary tract may be of different types such as, ureteropelvic junction obstruction, multicystic dysplastic kidneys, hypoplastic kidneys, exstrophy of the bladder etc., those of the female genital tract may be dysmorphic uterus, septate uterus, bicorporeal uterus, septate cervix, vaginal septum etc. and those of the male genital tract may be hypospadias, epispadias, cryptorchidism etc. The congenital anomalies of genitourinary system may be responsible for different types of reproductive problems. Hence, the present work has been carried out to study the congenital malformations of genitourinary system in perinates.

Methods: The study was carried out in all consecutive born perinates, total 7985 births (both live and still born), from 28th weeks of gestation to 7 days after birth, collected from the Department of Obstetrics and Gynaecology, Assam Medical College and Hospital, Dibrugarh, Assam, during 2014-16. After collection, the babies were examined and informations regarding congenital anomalies were noted in a pretested structured proforma, after taking written consents from the parents. Statistical calculations were done and the results were presented in tabular form.

Results: In the present study, out of 7985 births, nine cases showed congenital anomalies of genitourinary system (1.127/1000 live births). Out of these nine cases, three cases showed undescended testis (bilateral), two cases showed polycystic kidney (one bilateral and one unilateral), two cases showed incomplete duplication of the ureter (unilateral), one case showed penile duplication with accessory perineal scrotum and one case showed exstrophy of the urinary bladder.

Conclusion: The incidence of congenital malformations, in the present study, was comparatively lower than the previous studies, which may help in the management of these anomalies.

KEYWORDS

congenital anomalies, genitourinary system, perinate

INTRODUCTION

Congenital anomalies of the genitourinary system are found in approximately 10% of infants[1]. Congenital anomalies of kidney and urinary tract (CAKUT) occur in 1/500 births[2]. Ureteropelvic junction obstruction is the most common congenital anomaly of kidney and urinary tract, responsible for an abdominal mass in a neonate, followed by multicystic dysplastic kidneys, hypoplastic kidneys, vesicoureteral reflux, nonobstructed nonrefluxing primary megaureter, and bladder outlet obstruction (e.g., posterior urethral valve)[3]. Other anomaly of the urinary tract includes exstrophy of the urinary bladder, where the lateral body wall folds fail to fuse in the midline in the pelvic region, resulting in a ventral body wall defect, exposing the bladder mucosa[4].

Congenital anomalies of the female genital tract are prevalent in approximately 4-7% of population. These develop as a result of embryological maldevelopment of the Müllerian or paramesonephric ducts[5]. According to ESHRE/ESGE classification system, congenital anomalies of uterus are classified into U0, normal uterus; U1, dysmorphic uterus; U2, septate uterus; U3, bicorporeal uterus; U4, hemiuterus; U5, aplastic uterus; U6, for still unclassified cases. Coexistent cervical anomalies are classified into C0, normal cervix; C1, septate cervix; C2, double cervix; C3, unilateral cervical aplasia; C4, cervical aplasia. Coexistent vaginal anomalies are classified into V0, normal vagina; V1, longitudinal non-obstructing vaginal septum; V2, longitudinal obstructing vaginal septum; V3, transverse vaginal septum and/or imperforate hymen; V4, vaginal aplasia[6].

Congenital anomalies of the male genital tract may be of different types, such as hypospadias (3-5/1000 births), in which there is incomplete fusion of the urethral folds and presence of abnormal openings of the urethra along the inferior aspect of the penis; epispadias (1/30,000 births), in which the urethral meatus is found on the dorsum of the penis; bifid penis or double penis, in which the genital tubercle splits; congenital indirect inguinal hernia, in which the processus vaginalis remains in open communication with the peritoneal cavity and cryptorchidism, in which one or both testes fail to descend to the scrotum[4].

Congenital anomalies of kidney and urinary tract (CAKUT) account for the most cases of pediatric end stage renal disease and predispose the individual to hypertension and cardiovascular disease[2]. Congenital anomalies of the female genital tract are relatively common benign condition. They are associated with different health and reproductive problems depending on the type and degree of anatomical distortion[7]. Similarly, the male genital tract anomalies may also be associated with different types of health and reproductive problems.

Hence, the present work has been carried out to study the congenital anomalies of genitourinary system in perinates.

MATERIALS AND METHODS

The present study was carried out in the Department of Anatomy, Assam Medical College & hospital, Dibrugarh, Assam. All consecutive born perinates, total 7985 births (both live and still born), from 28th weeks of gestation to 7 days after birth, were collected from the Department of Obstetrics and Gynaecology, Assam Medical College and Hospital, during 2014-16, after obtaining ethical clearance from the Institutional Ethical Committee. The fetuses born less than 28 weeks, abortus and macerated fetuses were not included in the study. After collection, the babies were examined and epidemiologic information were collected from the parents in a pretested structured proforma, prepared as per EUROCAT[8], with local modifications. The autopsy of the still born fetuses (after fulfillment of all official procedures) was done in the Department of Anatomy to detect presence of congenital malformations, and the expert opinion was sought for confirming diagnosis. When malformation was detected, information regarding birth order, sex, birth weight, h/o consanguinity, h/o teratogenic drug exposure was obtained by systematic maternal and paternal interviews and was noted in the proforma. Statistical calculations were done in Microsoft Excel sheets and the results were presented in tabular form.

RESULTS AND OBSERVATIONS

Out of total 7985 births, 7807 babies were live born and 178 were stillborn. Among this 4144 were male and 3841 were female. No cases with ambiguous ganitalia were found. Out of total 7985 births, nine cases showed congenital anomalies of genitourinary system (1.127/1000 live births). Out of these nine cases, three cases showed undescended testis (bilateral), two cases showed polycystic kidney (one bilateral and one unilateral), two cases showed incomplete duplication of the ureter (unilateral), one case showed penile duplication with accessory perineal scrotum and one case showed exstrophy of the urinary bladder. Out of two cases, one case of bilateral polycystic kidney showed associated anomaly of single umbilical artery (0.125/1000 live births). The types of the polycystic kidney were ascertained to be of childhood type due to relatively smooth external appearance of the kidneys. Out of nine cases of genitourinary system anomalies, five were found in females and four in males. Also, out of these nine cases, six babies were preterm and three babies were term.

Table 1: Incidences of congenital anomalies of genitourinary system

Congenital anomalies	Number of cases	Prevalence (per 1000 live births)
Undescended testis	03	0.724*
Polycystic kidney	02	0.25
Duplication of the ureter	02	0.25
Penile duplication with accessory perineal scrotum	01	0.241*
Bladder exstrophy	01	0.125

*per 1000 male live births

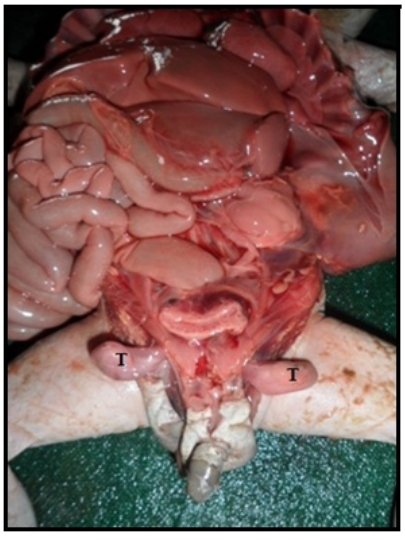

Figure 1: Photograph showing undescended testes (bilateral)

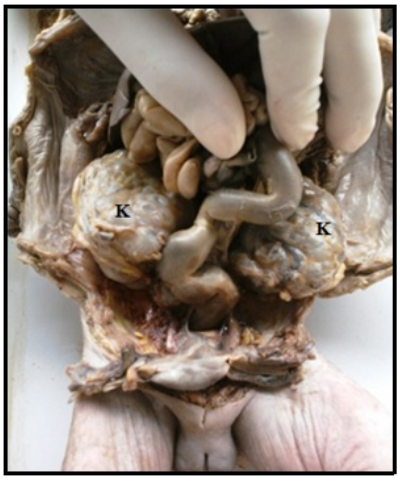

Figure 2: Photograph showing polycystic kidney (bilateral)

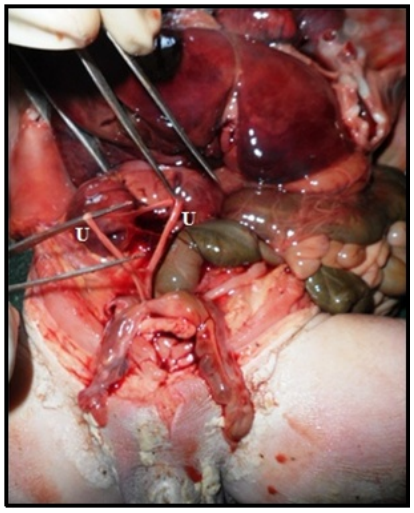

Figure 3: Photograph showing duplication of the ureter (unilateral)

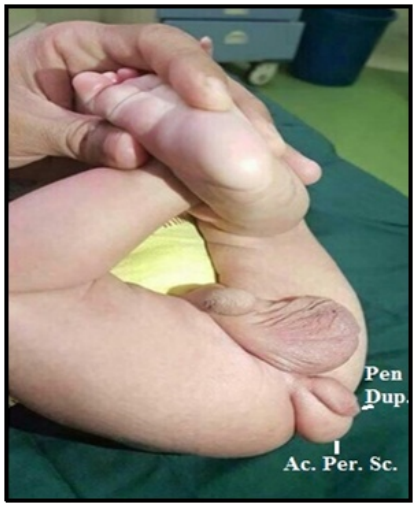

Figure 4: Photograph showing penile duplication with accessory perineal scrotum

Abbreviations: Key: K = Kidney; U = Ureter; T = Testis; Pen. Dup. = Penile Duplication; Ac. Per. Sc. = Accessory Perineal Scrotum

DISCUSSION

The incidence of genitourinary malformations of 1.127/1000 live births, in the present study, was comparable with Datta et al[9], who reported the rate as 1.02/1000 live births. McIntosh[10] with 7/1000, Ghosh et al[11] with 2/1000, Tibrewala et al[12] with 3.32/1000 live births have observed a higher incidence rate of genitourinary system malformations. Sugunabai et al[13] reported lower incidence rate of 0.6/1000 live births. The lowest incidence rate of 0.20/1000 live births was shown in the WHO sponsored study at Culcutta by Stevenson et al[14].

Abdullah et al[15] reported the incidence of undescended testes as 7.6/1000 male live births, Yucesan et al[16] reported the incidence as 9/1000 male live births and Thorup et al[17] reported the incidence as 20/1000 male live births. Pierik et al[18] reported the incidence of undescended testes at the median age of 35 as 12/1000 male live births. In the present study, the incidence of undescended testes was much less than the previous studies (0.724/1000 male live births).

Sadler[4] reported the incidence of childhood or autosomal recessive polycystic kidney disease (ARPKD) as 1/5,000 live births. Mandell et al[19] reported annual incidence of ARPKD as from 1 in 6000 to 1 in 14000 live births. According to Sessa et al[20], ARPKD occurs in 1/20,000 live births. Niaudet[21] mentioned the incidence of ARPKD to be 1/10,000 to 1/40,000 children per year. In the present study, the incidence of ARPKD was found to be 0.25 /1000, that is 1/4000 live births, which is similar to the findings of Sadler[4].

Das et al[22] mentioned the incidence of incomplete duplication of the ureter to be 0.4%. Nordmark[23] mentioned the incidence of unilateral incomplete division of the ureter to be 1.3%, unilateral complete division of the ureter to be 1.3%, bilateral complete division of the ureter to be 0.2%, bilateral incomplete division of the ureter to be 0.1% and complete division of the ureter on one side, and incomplete division of the ureter on the other side to be 0.06%. Timothy et al[24] mentioned the incidence of ureteral duplication to be 0.6% in autopsy cases. In the present study, unilateral incomplete duplication of the ureter was found to be 0.25/1000 live births, that is 0.025%, which is lower than the previous studies.

Melekos et al[25] reported a case of penile duplication in an eight year old boy, where the duplication was incomplete with a common root of the penis. Gyftopoulos et al[26] reported two cases of penile duplication, where one case showed true complete penile duplication associated with a bifid scrotum with undescended testes, while the other case showed true complete penile duplication, bladder and urethral duplication. Bakheet et al[27]reported a case of complete penile duplication with two scrotums and each one carrying two palpable testes. Both penises had normal shaft with normally located meati. Elsawy[28] reported a case of penile duplication with two penises, one dorsally and one ventrally located.

Tripathi et al[29] reported a case, where a 11 months old male child presented with a small accessory scrotum attached to a well developed 'primary' scrotum, which was completely normal containing two normal testes. The accessory scrotal sac was empty. The penis was normally developed. Ito et al[30] reported a case, where there was a coexistence of midperineal and lateral types of accessory scrotum. Chadha et al[31] reported a case with a small phallus, bifid scrotum and absence of the scrotal raphe. Tirtayasa et al[32] reported a case with complete bifid diphallia associated with bifid scrotum. Kode[33] mentioned that the extent of penile duplication ranges from a small accessory penis anywhere on the normal penis, bifid penises, to a complete duplication of the penis, scrotum, testes, bladder and urethra. Adair et al[34] mentioned that the accessory penis may be present anywhere on the penile shaft or perineum. In the present study, a newborn presented with an accessory penis and an accessory scrotum in the perineum along with the normal penis and normal scrotum, both the scrotums containing the testes with no abnormality of the meati and any other associated anomaly, which was not reported in the literature earlier.

Sadler[4] reported the incidence of bladder exstrophy as 2/10,000 live births, Gross et al[35] mentioned the incidence as 1/50,000 live births, Nelson et al[36]reported the incidence as 2.15/100,000 live births, Caton et al[37] reported the incidence as 2.1/100,000 live births and Siffel et al[38] mentioned the incidence as 2.07/100,000 live births. In the present study, the incidence of bladder exstrophy was 0.125/1000 live births, that is 1.25/10,000 live births, which is more similar to the findings of Sadler[4].

CONCLUSION

In conclusion, the incidence of congenital malformations, in the present study, was comparatively lower than the studies in other parts of India and abroad. The present study has helped us to know the pattern and frequency of congenital malformations of genitourinary system and the associated anomalies, which will help us to plan future strategies for prevention, early diagnosis and timely management of such cases.

Funding: None
Conflict of interest: None declared

REFERENCES

1. Cohen HL, Kravets F, Zucconi W, Ratani R, Shah S, Dougherty D. Congenital abnormalities of the genitourinary system. Semin Roentgenol. 2004; 39(2):282-303.
2. Song R, Yosypiv IV. Genetics of congenital anomalies of the kidney and urinary tract. Pediatr Nephrol. 2011; 26(3):353-64.
3. Pope JC, Brock JW, Adams MC, Stephens FD, Ichikawa I. How they begin and how they end: classic and new theories for the development and deterioration of congenital anomalies of the kidney and urinary tract, CAKUT. J Am Soc Nephrol. 1999; 10(9):2018-28.
4. Sadler TW. Urogenital system. In: Langman's Medical Embryology. 13th ed. Wolters Kluwer. Philadelphia, 2015:255-76.
5. Chan YY, Jayaprakasan K, Zamora J, Thornton JG, Raine-Fenning N, Coomarasamy A. The prevalence of congenital uterine anomalies in unselected and high-risk populations: a systematic review. Hum Reprod Update. 2011; 17(6):761- 71.
6. Grimbizis GF, Gordts S, Spiezio Sardo AD, Brucker S, Angelis CD, Gergolet M, Li TC, Tanos V, Brolmann H, Rudi Campo LG. The ESHRE/ESGE consensus on the classification of female genital tract congenital anomalies. Hum Reprod. 2013; 28(8):2032-44.
7. Gergolet M, Campo R, Verdenik I, KendaSuster N, Gordts S, Gianaroli L. No clinical relevance of the height of fundal indentation in subseptate or arcuate uterus: a prospective study. Reprod Biomed Online. 2012; 24(5):576-82.
8. EUROCAT. EUROCAT Guide 1.3 and reference documents, instructions for the registration and surveillance of congenital anomalies, September 2005. Available at: http://www.eurocat-network.eu/content/EUROCAT-Guide-1.3.pdf
9. Datta V, Chaturvedi P. Congenital malformations in rural Maharastra. Ind Pediatr. 2000; 37(9):988-1001.
10. McIntosh R, Merritt KK, Richards MR, Samuels MH, Bellows MT. The incidence of congenital malformations: a study of 5964 pregnancies. Pediatrics. 1954; 14(5):505-22.
11. Ghosh S, Bali L. Congenital malformations in the newborn. Indian J Child Health.1963; 12:448-52.
12. Tibrewala NS, Pai PM. Congenital malformations in the newborn period. Ind Pediatr. 1974; 11(6):403-06.
13. Suguna Bai NS, Mascarene M, Syamalan K, Nair PM. An etiological study of congenital malformation in the new born. Ind Pediatr. 1982; 19(12):1003-7.
14. Stevenson AC, Johnston HA, Stewart MI, Golding DR. Congenital malformations: a report of study of series of consecutive births in 24 centers. Bull World Health Organ. 1966; 34(Suppl):9–127.
15. Abdullah NA, Pearce MS, Parker L, Wilkinson JR, Jaffray B, McNally RJQ. Birth prevalence of cryptorchidism and hypospadias in northern England, 1993-2000. Arch Dis Child. 2007; 92(7):565-67.
16. Yucasan S, Dindar H, Olcay I, Okur H, Kilicaslan S, Ergoren Y, Tuysuz C, Koca M, Civilo B, En IS. Prevalence of congenital abnormalities in Turkish school children. Eur J Epidemiol. 1993; 9(4):373-80.
17. Thorup J, Cortes D. The incidence of maldescended testes in Denmark. Peditr Surg Int. 1990; 5(1):2-5.
18. Pierik FH, Burdorf A, Sabine MPF, Keizer-Schrama DM, Wolffenbuttel KP, Rien Nijman JM, Jutttmann RE, Weber RFA. The cryptorchidism prevalence in infants in the general population of Rotterdam, the Netherlands. Int J Androl. 2005; 28(4):248-52.
19. Mandell J, Koch WK, Nidess R, Preminger GM, McFarland E. Congenital polycystic kidney disease: genetically transmitted infantile polycystic kidney disease in C57BL/6J mice. Am J Pathol. 1983; 113(1):112-14.
20. Sessa A, Meroni M, Righetti M, Battini G, Maglio A, Puricelli SL. Autosomal recessive polycystic kidney disease. Contrib Nephrol. 2001; 136:50-6.
21. Niaudet P. Autosomal recessive polycystic kidney disease. Orphanet encyclopedia. March 2004.
22. Das S, Dhar P, Mehra RD. Unilateral isolated bifid ureter: a case report. J Anat Soc India. 2001; 50(1):43-4.
23. Nordmark B. Double Formations of the pelves of the kidneys and the ureters: embryology, occurrence and clinical significance. Acta Radiologica. 1948; 30(11):267-78.
24. Timothy RP, Decter A, Perlmutter AD. Ureteral duplication: clinical findings and therapy in 46 children. The Journal of Urology. 1971; 105(3):445-51.
25. Melekos MD, Barbalias GA, Asbach HW. Penile duplication. Urology. 1986; 27(3):258-9.
26. Gyftopoulos K, Wolffenbuttel KP, Nijman RJ. Clinical and embryologic aspects of penile duplication and associated anomalies. Urology. 2002; 60(4):675-9.
27. Bakheet MA, Refaei M. Penile duplication and two anal openings: report of a very rare case. Iran J Pediatr. 2012; 22(1):133-6.
28. Elsawy M, Salle JLP, Abdulsalam M, Alsaid AN. Penile duplication: is it necessary to excise one of the penises? J Pediatr Urol. 2012; 8(4):434-6.
29. Tripathi FM, Khanna S. Accessory scrotum: a case report. Br J Plast Surg. 1983; 36:127-8.
30. Ito M, Hashizume K, Kanamori Y. New phenotype of accessory scrotum with perineal lipoblastoma: coexistence of midperineal and lateral accessory scrotums. Int J urol. 2004; 11(2):125-7.
31. Chadha R, Gupta S, Mahajan JK, Kothari SK. Two cases of pseudoduplication of the external genitalia. Pediatric Surg Int. 2001; 17(7):572-4.
32. Tirtayasa PMW, Prasetyo RB, Rodjani A. Diphallia with associated anomalies: a case report and literature review. Case Rep Urol. 2013; 2013. Available from:http://dx.doi.org/10.1155/2013/192960
33. Kode GM. Penile duplication. Br J Plast Surg. 1991; 44:151-2.
34. Adair EL, Lewis EL. Ectopic scrotum and diphallia: a case report. Journal of Urology. 1960; 84:115.
35. Gross RE, Cresson SL. Exstrophy of bladder: observation from eighty cases. JAMA. 1952; 149(18):1640-4.
36. Nelson CP, Dunn RL, Wei JT. Contemporary epidemiology of bladder exstrophy in the United States. The Journal of Urology. 2005; 173(5):1728-31.
37. Caton AR, Bloom A, Druschel CM, Kirby RS. Epidemiology of bladder and cloacal exstophies in New York state, 1983-1999. Birth Defects Res A Clin Mol Teratol. 2007; 79(11):781-7.
38. Siffel C, Correa A, Amar E, Bakker MK, Barmejo-Sanchez E, Bianca S, Castilla EE, Clementi M et al. Bladder exstrophy: an epidemiologic study from the International clearinghouse for birth defects surveillence and research, an overview of the literature. Am J Med Genet C Semin Med Genet. 2011; 157(4):321-332.