

A Survey of Externally Recognizable Genitourinary Anomalies in Korean Newborns*

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To estimate the incidence of externally recognizable genitourinary anomalies and associated anomalies in the newborns in Korea, retrospective and prospective studies have been performed. Thirty eight of 48 urology training hospitals participated in this nationwide survey. In this study we have included minor defects or variations in the anomaly to evaluate the incidence of the recognizable genitourinary conditions in the newborns.

The incidence of genitourinary anomaly in 1,000 newborn delivery in a year was 11.0 in the prospective study and this figure is about three times higher than the retrospective study and is considered to be close to the true incidence. The incidence of genitourinary anomaly in 1,000 male newborn was 20.4. Hydrocele, cryptorchidism and hypospadias were most commonly observed. The incidence of hydrocele in 1,000 male newborn was 9.89 and the incidence of cryptorchidism was 7.26 and the incidence of hypospadias was 2.13. The incidence of associated anomaly in 100 genitourinary anomaly was 7.9. Congenital heart diseases and anorectal anomalies were commonly associated anomalies. In the newborns with genitourinary anomalies, premature infants account larger portion than is usually reported in total delivery.

Key Words: *Incidence of genitourinary anomalies, newborns*

INTRODUCTION

Recently the incidence of the genitourinary anomalies in newborns is becoming more prevalent with the development of the pediatric urology. There have been some reports on the prevalence of congenital anomalies in Korean newborns, but most of the studies were retrospective review based on hospital medical records (Ahn, 1975; Choi et al., 1985; Jeon et al., 1987). The incidences of the above stu-

dies were much lower than the incidences of the western countries and might not reflect the true figures.

Both retrospective and prospective studies on the incidence of externally recognizable genitourinary anomalies were performed in participation of 38 training hospitals by the Korean Urological Association. The prospective study was designed to have the newborns examined at the nursery room by a urologist. Minor defects or normal variations are included in the anomaly to evaluate the figures of all the recognizable genitourinary diseases in the newborns and to compare the results with previous reports. And examination of other organ anomalies associated with genitourinary anomaly is thought to be important to take care of the anomalous patients. The results of this study can also be helpful in estimating the proper manpower needed in pediatric urology in

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Korea.

MATERIALS AND METHODS

The survey has been performed in 28 university hospitals and 10 large training hospitals in Korea. Twenty three hospitals are located in Seoul, four in Pusan, four in Taegu, two in Kwangjoo, one in Choongnam Province, one in Kangwon Province, one in Chunbook Province and two in Kyungnam Province.

The study was divided into two parts, retrospective study and prospective study. In retrospective study, the data were collected from newborn records of babies born in one year period (from January 1 to December 31 in 1986). In prospective study, a participating urologist visited nursery room and examined newborns on the first or second day after birth during 5 months period (from November 1 to December 31 in 1987 and from March 1 to May 31 in 1988). The examiner checked and collected the patient's data on genitourinary anomalies and associated anomalies in newborns. Using the formal examination sheet, the examiner checked name, sex, birth date, gestational period, birth weight, parity and delivery type in every newborn baby with the externally recognizable genitourinary anomalies. In retrospective study, data only from the patients with the diagnosis of ambiguous genitalia, hypospadias, cryptorchidism, hydrocele and abdominal mass were collected and analysed. But in prospective study, we used check list in order that the examiner might not miss to examine any part and to report any finding.

The content of the check list was as follows:

1. The number of umbilical artery: one (single), two
2. Abdominal mass: yes, no
3. Prepuce: phimosis, redundant prepuce, abnormal prepuce
4. Shaft of penis: straight, curved
5. Position of meatus: normal, coronal, penile, penoscrotal, scrotal or perineal, epispadias
6. Shape of the scrotum: relaxed, retracted, enlarged and transilluminated, enlarged and not transilluminated, bifid scrotum
7. Position of testis: normal, upper scrotal, inguinal, impalpable, in scrotum but hard and enlarged

All examination papers also contained check list for associated anomalies: Down's syndrome, hydrocephaly, ear malformation, cleft lip, cleft palate, congenital heart disease, myelodysplasia, imperforate anus, polydactyly and syndactyly. Other anomalies found were asked to be written in the provided space. These associated anomalies were usually detected by a pediatrician of the nursery of the participating

hospital.

RESULT

The number of total newborns in nurseries during one year of 1986 in the 38 participating hospitals of the retrospective study was 21,806 (male: female = 11,161:10,645). The number of total newborns during 5 months of 1987 and 1988 in the prospective study was 14,854 (male: female = 7,990:6,864). The total population in this study consisted 36,660 newborns and male: female ratio was 52.2:47.8.

1. Incidence of genitourinary anomalies and associated anomalies

In the prospective study (5 months of 1987 and 1988), genitourinary anomalies were observed in a total of 164: male 163 and female 1. Incidence of genitourinary anomaly during this period was calculated to be 11.0 per 1,000 newborn delivery per year and 20.4 per 1,000 male newborn per year. However in the retrospective study (in year of 1986), genitourinary anomalies were found in a total of 82 newborns: male 77 and female 5. Incidence of genitourinary anomalies during this period was 3.8 per 1,000 newborn delivery per year and 6.8 per 1,000 male newborn per year (Table 1).

The incidence of associated anomalies in other organ with genitourinary anomalies was 7.9 per 100 genitourinary anomalous newborn in the prospective study. In retrospective study, the incidence was 18.3 (Table 2).

2. Distribution and relative frequency of each genitourinary anomaly

In the prospective study, the most common genitourinary anomaly was hydrocele (41.8%), followed by cryptorchidism (30.7%), hypospadias (9.0%), inguinal hernia (5.8%), abdominal mass (3.2%), ambiguous genitalia (1.1%), micropenis (1.1%), epispadias (1.1%) and exstrophy (1.1%) (Table 3). To estimate more accurate incidence of each genitourinary anomaly in newborn, we analyzed the data of the prospective study. The incidence of hydrocele in 1,000 male newborn was 9.89 and that of cryptorchism was 7.26, hypospadias 2.13, inguinal hernia 1.38, and abdominal mass 0.75 (Table 4).

As the informations collected from the patients with genitourinary anomalies are same in the prospective and retrospective studies, analysis of the associated anomalies and comparison of epidemiologic factors were made from the combined data of both prospective and retrospective studies.

Multiple genitourinary anomalies were observed in 30 newborns and its frequency was 12.2 percent in

Table 1. Incidence of newborn genitourinary anomalies

	Retrospective 1986	Prospective 1987-1988
Total No. of delivery	21,806	14,854
Total No. of male newborn	11,161	7,990
No. of newborn GU anomaly	82 (male: 77)	164 (male: 163)
Incidence /1,000 delivery/yr (95% C.I. *)	3.8 (1.4-10.0)	11.0 (6.1-19.7)
Incidence /1,000 male newborn /yr. (95% C.I. *)	6.8 (3.3-14.2)	20.4 (13.3-31.4)

*Confidence interval was estimated by analysis of a single rate using asymptotic model.

Table 2. Proportion of associated anomalies among newborns with genitourinary anomalies

	Retrospective 1986	Prospective 1987-1988
No. of newborn with GU anomaly	82	164
No. of newborn with associated anomaly	15	13
Proportion of associated anomaly	18.3%	7.9%

Table 3. Relative frequency of newborn genitourinary anomaly

Anomalies	Retrospective 1986 No.	Prospective 1987-1988 No. (%)
Hydrocele	23	79 (41.8)
Cryptorchism	28	58 (30.7)
Hypospadias	9	17 (9.0)
Inguinal hernia	4	11 (5.8)
Abdominal mass	4	6 (3.2)
Ambiguous genitalia	2	2 (1.1)
Micropenis	2	2 (1.1)
Epispadias	0	2 (1.1)
Exstrophy	0	2 (1.1)
Potter's synd (Agenesis of kidney)	0	1 (0.5)
Testicular torsion	0	2 (1.1)
Prune-belly syndrome	1	0 (0)
Others	6	7 (3.7)
Total	89	189 (100)

a total of 246 genitourinary anomalous newborns. The number of newborns with having both cryptorchidism and hydrocele was 10 (33.3%) and both cryptorchidism and hypospadias was 4 (13.3%).

3. Relative frequency of associated anomalies with genitourinary anomalies

Forty three anomalies of other organs were found in 28 genitourinary anomalous newborns. The most commonly associated anomaly was congenital heart disease (16.3%) and the next were imperforate anus (14.0%), equinovarus (7.0%), tongue tie (7.0%), Down's syndrome (7.0%) and anencephaly (4.7%) in order of

Table 4. Incidence of various genitourinary anomalies

Anomalies	Incidence/1,000 male newborn
Hydrocele	9.89
Cryptorchism	7.26
Hypospadias	2.13
Inguinal hernia	1.38
Abdominal mass	0.75 (0.40*)

*Incidence/1,000 total delivery

Table 5. Associated anomalies of other systems

Associated anomalies	No. (%)
Congenital heart disease	7 (16.3)
Imperforate anus	6 (14.0)
Equinovarus	3 (7.0)
Tongue tie	3 (7.0)
Down's syndrome	3 (7.0)
Anencephaly	2 (4.7)
Others	19 (44.2)
Total	43 (100)

Table 6. Associated anomalies classified by system

System	No. (%)
Gastrointestinal	14 (32.6)
Musculoskeletal	10 (23.3)
Central nervous	7 (16.3)
Cardiovascular	7 (16.3)
Skin	4 (9.3)
Respiratory	7 (2.3)
Others	1 (2.3)
Total	43 (100)

frequency. Others were microcephaly, hypertelorism, malformed ear, low set ear, mandibular hypoplasia, cleft lip, cleft palate, pneumothorax, intestinal obstruction, malrotation of bowel, gastroschisis, club foot, polydactyly, syndactyly and etc. (Table 5). When classifying these associated anomalies by system, the most common one was gastrointestinal (32.6%) and the next were musculoskeletal (23.3%), central nervous (16.3%), cardiovascular (16.3%), skin (9.3%), respiratory (2.3%) and others (2.3%) in order of frequency (Table 6).

4. Comparison of genitourinary anomalies with gestational period, birth weight, parity and delivery type

Among 246 newborns with genitourinary anomaly, 234 newborn records were adequate for analysis. In these 234 newborns there were 276 genitourinary anomalies. By gestational period, 234 newborns were grouped as those less than 37 weeks was 40 (17.1%), 37 weeks or more 183 (78.2%) and unchecked 11

(4.7%). By birth weight, 2500gm or less was (17.1%), greater than 2,500gm 189 (80.8%) and unchecked 5 (2.1%). By parity, primipara was 92 (39.3%), multipara 123 (52.6%) and unchecked 19 (8.1%). By delivery type, cesarean section was 57 (24.4%), vaginal delivery 166 (70.9%) and unchecked 11 (4.7%) (Table 7).

We chose five most frequent anomalies such as hydrocele, cryptorchidism, hypospadias, inguinal hernia and abdominal mass and then we observed the distribution of these anomalies by above parameters (Table 8, 9, 10). In gestational period and birth weight, the data was compared with the proportion of premature infants in a total delivery of Seoul National University Hospital during 7 years (1980-1986). The percentage of prematurity among genitourinary anomalies, judged by gestational age and weight, was significantly higher than the one in a total delivery ($p < 0.01$).

Table 7. Gestational period, birth weight, parity and delivery type of 234 newborns with genitourinary anomaly

	No. (%)
Gestational period	
less than 37 weeks	40 (17.1)
37 weeks or more	183 (78.2)
unchecked	11 (4.7)
Birth weight	
2,500gm or less	40 (17.1)
more than 2,500gm	189 (80.8)
unchecked	5 (2.1)
Parity	
primipara	85 (36.3)
multipara	97 (41.5)
unchecked	52 (22.2)
Delivery type	
cesarean section	57 (24.4)
vaginal delivery	166 (70.9)
unchecked	11 (4.7)

Table 8. Comparison of various genitourinary anomalies with gestational period

Anomaly	Gestational period			Total No.	z*	p-value
	<37wks No. (%)	≥37wks No. (%)	Unknown No. (%)			
Hydrocele	11 (10.2)	93 (86.1)	4 (3.7)	108	-0.806	N.S.
Cryptorchism	19 (23.2)	62 (75.6)	1 (1.2)	82	-3.279	**P<0.01
Hypospadias	5 (18.5)	20 (74.1)	2 (7.4)	27	-1.377	N.S.
Inguinal hernia	4 (28.6)	7 (50.0)	3 (21.4)	14	-1.526	N.S.
Abdominal mass	1 (12.5)	7 (87.5)	0 (0)	8	-0.402	N.S.
Others	10 (27.0)	25 (67.6)	2 (5.4)	37	—	—
Total	50 (18.1)	214 (77.5)	12 (4.3)	276	-4.323	p<0.01

*Statistical significances of the proportion of newborn under 37 weeks between anomalous and normal newborns in Seoul National University Hospital, 1980-1986 (n = 11,770, p = 0.078) was calculated by the analysis of proportion using asymptotic z-distribution.

**p<0.01; highly significant.

N.S.; non-significant.

Table 9. Comparison of various genitourinary anomalies with birth weight

Anomaly	Birth weight			Total No.	z*	p-value
	≤2500gm No. (%)	>2500gm No. (%)	Unknown No. (%)			
Hydrocele	10 (9.3)	96 (88.9)	2 (1.9)	108	-0.141	N.S.
Cryptorchism	20 (24.4)	61 (74.4)	1 (1.2)	82	-3.243	**P<0.01
Hypospadias	5 (18.5)	21 (77.8)	1 (3.7)	27	-1.260	N.S.
Inguinal hernia	3 (21.4)	9 (64.3)	2 (14.3)	14	-1.056	N.S.
Abdominal mass	2 (25.0)	6 (75.0)	0 (0)	8	-1.051	N.S.
Others	9 (24.3)	28 (75.7)	0 (0)	37	—	—
Total	49 (17.8)	221 (80.1)	6 (2.2)	276	-3.799	p<0.01

*Statistical significances of the proportion of newborn under 2,500gm between anomalous and normal newborns in Seoul National University Hospital, 1980-1986 (n = 11,770, p = 0.089) was calculated by the analysis of proportion using asymptotic z-distribution.

**p<0.01; highly significant.

N. S.; non-significant.

Table 10. Comparison of various genitourinary anomalies with parity

Anomaly	Parity			Total No.
	Nullipara No. (%)	Multipara No. (%)	Unknown No. (%)	
Hydrocele	40 (37.0)	50 (46.3)	18 (16.7)	108
Cryptorchism	26 (31.7)	30 (36.6)	26 (31.7)	82
Hypospadias	7 (25.9)	14 (51.9)	6 (22.2)	27
Inguinal hernia	5 (35.7)	4 (28.6)	5 (35.7)	14
Abdominal mass	3 (37.5)	5 (62.5)	0 (0)	8
Others	16 (43.2)	12 (32.4)	9 (24.3)	37
Total	97 (35.1)	115 (41.7)	64 (23.2)	276

Table 11. Location and laterality of cryptorchid testes

Laterality	Upper scrotal No. (%)	Location Inguinal No. (%)	Impalpable No. (%)	Total No. (%)
Unilateral	9 (30.0)	11 (36.7)	10 (33.3)	30 (100)
Rt.	5	3	4	12
Lt.	4	8	6	18
Bilateral	16 (28.6)	24 (42.9)	16 (28.6)	56 (100)
symmetric	12	18	14	44
asymmetric	4	6	2	12
Total	25 (29.1)	35 (40.7)	26 (30.2)	86 (100)

5. Other clinical data in the prospective study

1) Number of umbilical artery in genitourinary anomaly cases

The data on the number of umbilical artery were available in 148 of total 164 genitourinary anomalous newborns. Single umbilical artery was found in 18 (12.2%) and two umbilical arteries were found in 130 (87.8%). Single umbilical artery was found in 15.8% of cryptorchidism, 8.6% of hydrocele and 5.9% of hypospadias. Also, each one case of micropenis, exstrophy and bilateral renal agenesis had single umbilical artery.

2) Hydrocele

In 79 cases of hydrocele, the involved side was recorded in 74 cases. Left was 14 (18.9%), right 35 (47.3%) and bilateral 25 (33.8%).

3) Hypospadias

In 17 cases of hypospadias, coronal hypospadias was 6 (35.3%), penile 10 (58.8%) and penoscrotal 1 (5.9%).

4) Cryptorchidism

In 58 cases of cryptorchidism, 30 cases (51.7%) were unilateral and 28 cases (48.3%) were bilateral. In 30 patients of unilateral undescended testis, 12 (40%) were on the right side while 18 (60%) were

on the left. In 22 (78.6%) of the 28 bilateral undescended testes both testes were found in the same anatomical location. When we classified the position of cryptorchid testes by upper scrotal testis, inguinal testis and impalpable testis, a total of 86 cryptorchid testes in 58 cryptorchid newborns could be divided as follows; upper scrotal testis was 25 (29.1%), inguinal testis 35 (40.7%) and impalpable testis 23 (30.2%) (Table 11).

5) Abdominal Mass

Abdominal mass was found in 6 cases (male 5 and female 1). Etiological classification revealed hydronephrosis in 3 cases (50%), adrenal pyocyst 1 case (16.7%) and unknown 2 cases (33.3%).

DISCUSSION

Congenital malformation is a structural anomaly which appears in newborns of any causes and it excludes the anomalies that appear a few months or years after delivery (Warkany, 1971). In this study, genitourinary anomaly represents structural malformation including minor defects and variations which is recognizable externally in newborn.

There have been several studies to define the incidences of congenital anomalies (McIntosh et al., 1954; Hendricks, 1955; Marden et al., 1964; Warkany, 1971). It is difficult to compare the incidences of various series because the criteria of anomalies were different from study to study. Anomaly can be divided to major and minor: a major anomaly is one which has an adverse effect on either the function or social acceptability of the individual and a minor anomaly is one which has neither of medical nor cosmetic consequence to the patient. Most studies are concerned with the incidence of major anomalies and about 2% of newborn infants have a major anomaly. If one includes anomalies detected at subsequent ages the incidence is as high as 7.5% in western countries (McIntosh et al., 1954). A survey of over 4,000 newborn babies by surface examination revealed that 14.7% had at least one minor anomaly and suggested the use of minor anomaly as indicators of altered embryonic differentiation (Marden et al., 1964).

On several reports in Korea, the range of frequency of congenital malformations was 0.77-6.8% (average 3.18%) and the anomalies of genitourinary organ system occupied about 14-19% of the total newborn anomalies (Ahn, 1975; Choi et al., 1985; Jeon et al., 1987). These studies were retrospective review of hospital medical records and followed the McIntosh's classification of congenital malformation and included the hydrocele and inguinal hernia. World Health Organization excluded the congenital hydrocele and inguinal hernia from the congenital anomaly section. But here in this study we have included because our interest were not only in the genitourinary anomaly in a narrow sense but also in the recognizable genitourinary diseases or conditions in the Korean newborns. The frequency of genitourinary malformations can be calculated from the above series; 1.39% in Ahn's study (1975), 0.42% in Choi's study (1985) and 0.46% in Jeon's study (1987). In the present study, the frequency of genitourinary anomalies was 1.1%. The genitourinary anomalies in newborn were more frequently found in the prospective study than the retrospective one. The low incidence of retrospective study is presumably due to incomplete examination or recording. Many minor anomalies or even major one might have been overlooked by the physician. Examination of the newborn with suspected genitourinary anomaly will yield a reliable data if the examiners use a check list which will help to keep the criteria of anomaly constant.

In our study, hydrocele was the most prevalent

genitourinary anomaly and the second was cryptorchidism. Ahn (1975) and Choi et al. (1985) reported that hydrocele were found more frequently than cryptorchidism but Lee et al. (1983) and Jeon et al. (1987) reported that cryptorchidism was found more frequently. Generally it was reported that hydrocele was observed in 6% among term babies (Scorer and Farrington, 1971) and cryptorchidism was observed in 3.4% of term baby, 30.3% of premature baby and 4.3% of total delivery (Scorer, 1964) and hypospadias was observed in 0.82% of total delivery (Sweet et al., 1974). Lee et al. (1983) reported that the relative frequency of hydrocele was 2.8%, cryptorchidism 4.5%, hypospadias 0.2% but in our study these were 0.99%, 0.73% and 0.21% respectively. Hydroceles in the newborn period are physiologic and the difference in incidence might be due to different criteria. But the incidence of cryptorchidism might be truly lower in our country because in this newborn period there is no cremasteric reflex and a little chance of misdiagnosis. In those rare cases like epispadias, exstrophy, prune-belly syndrome and myelodysplasia the incidence could not be estimated because of this small epidemiologic sample size.

Multiple genitourinary anomalies in a newborn was observed in 30 newborns (12.2%). The association of anomalies in other organ systems was found in 9.7%. Congenital heart disease and imperforate anus were observed as frequent associated anomalies. It is known that the incidence of anomalies of the urinary tract is high in the patients of congenital heart disease. On excretory urography after cardiac catheterization renal anomalies were found in 10.8% (King, 1969) and in 3.6% in Korea (Lee et al., 1989). One fourth to over one half of all cases of imperforate anus had associated anomalies of the genitourinary system other than the fistula and 28% were reported in Korea (Jung and Cho, 1987).

Though the percentage of prematurity among genitourinary anomalies was significantly higher than usual incidence in a total delivery, we cannot say that all of the genitourinary anomaly occur more frequently in premature babies. But cryptorchidism seemed to be occurred more commonly in premature babies. Embryologically descending of testis occurs in the last two months of fetal life and it is well known the cryptorchidism is very frequently found in premature babies.

Hendricks (1955) and Ahn (1975) reported that occurrence of a congenital anomaly was not related with parity. Lee et al. (1980) also reported that occurrence of a genitourinary anomaly was not related

with parity. We could not identify the relationship of anomaly with parity too.

Bilateral cryptorchidism was observed in 48.3% of all cryptorchid newborns in our study, which was somewhat higher than usual incidence, 10-30% of clinical study in children (Belman et al., 1988). But our results are similar to Villumsen's 56.5% (1966) and Lee's 53.4% (1983) in their newborn studies. This higher incidence may be explained by that majority of this bilateral cryptorchid testes descend during 3 months after birth by hormonal effect and after 6 months of age the incidence of bilateral cryptorchidism decreases to the expected incidence in children (Scorer, 1956; Frey et al., 1982). About 75% of testes undescended at birth were in normal position by the age of one year (Villumsen and Zachau-Christiansen, 1966). In our series 29.1% of the undescended testes could reach upper scrotum on traction and this group might be the one that can be expected to descend spontaneously. The proportion of impalpable testis in newborn cryptorchid testes was 30.2% in our study. Buemann et al. (1961) reported the incidence was 67.3% and Lee et al. (1983) 10.2%. Levitt et al. (1978) and Belman et al. (1988) reported the proportion of impalpable testis in operative cases was 20-24%. Buemann's (1961) high incidence can be explained by the high incidence of prematures in his study.

CONCLUSION

To estimate the incidence of genitourinary anomalies in newborns in Korea, retrospective and prospective studies were undergone in urology training hospitals.

The incidence of genitourinary anomalies in 1,000 newborn deliveries in a year was 11.0 in the prospective study, about three times higher than the one in the retrospective study. The incidence of genitourinary anomalies in 1,000 male newborns in a year was 20.4. Among genitourinary anomalies, hydrocele was the most common one (41.8%), the second cryptorchidism (30.7%) and the third hypospadias (9.0%). The incidences of these anomalies in 1,000 male newborn were 9.89, 7.26 and 2.13 respectively. The incidence of associated anomaly with 100 genitourinary was 7.9. Congenital heart disease (16.3%) and imperforate anus (14.0%) were commonly associated anomalies with a genitourinary anomaly. The percentage of prematurity among genitourinary anomalies, judged by gestational age and birth weight, was significantly higher than the one in a total delivery.

Of the cryptorchidism newborns, 48.3% were bilateral. Testis were located in upper scrotum in 29.1%, inguinal canal in 40.7% and were impalpable in 30.2%.

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