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Major urogenital malformations in Nigerian children

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Abstract:

Background

There is scanty data on the pattern of childhood urogenital malformations in many resource-poor countries, including Nigeria.

Objective

To describe the pattern of occurrence of major urogenital malformations in children presenting in a tertiary health care centre in sub Saharan Africa.

Design

Retrospective case review.

Patients

All children with major urogenital malformations presented at the Department of Paediatrics of the University College Hospital, Ibadan, a large tertiary health care institution in South West Nigeria. In the period from July 1985 to December 1995.

Main results

A total number of 125 cases of major urogenital malformations were seen during the study period. The commonest of these malformations were posterior urethral valves (40.0%), hypospedias (18.4%), ambiguous external genitalia (12.8%) and ectopia vesicae (11.2%).
Prenatal diagnosis was rare and most patients presented late. Mortality during first admission was 14.4% (18/125), mostly accounted for by cases of posterior urethral valves.

Conclusion

Posterior urethral valves accounted for the majority of major urogenital malformations seen in the setting studied.
The high mortality probably reflected the severity of the lesions and late presentation of patients (which was related to the rarity of prenatal diagnosis). Increased awareness of these lesions among physicians practising in developing countries and greater use of ultrasonography in the third trimester of pregnancy may improve the prognosis by early detection and management.

Key words: epidemiology; prenatal diagnosis; urogenital system abnormalities

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Introduction

Malformations of the urogenital system are not uncommon. Prenatal diagnosis by fetal ultrasonography has provided an insight into the magnitude of the problem and a means of early detection and intervention [1,3]. Early detection and management, reduces the risk of renal damage and chronic renal failure. This is especially important in developing countries where facilities for renal replacement therapy in childhood are scarce.
There is limited data on major urogenital malformations in Africa, including Nigeria. While there are many case reports and some series on specific conditions, few reports have examined the pattern of occurrence of these malformations. Incidence studies in the newborn was limited by the fact that only externally recognizable malformations such as hypospedias and cryptorchidism were recorded [4-6], leaving the vast majority of urinary tract lesions (such

as posterior urethral valves) unrepresented. Autopsy studies [7-8] are more useful but their validity rested on autopsy rates which are often low in developing countries. Moreover, many such malformations are not lethal and are therefore under-represented in autopsy studies. Generally, these studies provided an idea on the magnitude of the problem, but inadequate data on the relative occurrence of the various types of urogenital malformations and their role in morbidity which is needed for health planning purposes.

This retrospective study was undertaken to describe the pattern of urogenital malformations among children admitted into a major tertiary health institution in West Africa over a ten and half years period and to present data on short term outcome in such children.

Methods

This is a retrospective study of records from children aged 0-14 years presenting at the Department of Paediatrics, University College Hospital, Ibadan, Nigeria with a diagnosis of a major urogenital malformation over a 10 and a half year period (July 1, 1985 to December 31, 1995). A major urogenital malformation was defined as a congenital malformation in the urogenital system that has major medical, surgical or cosmetic consequences [9]. The definitive diagnosis in each case was made by clinical examination and imaging of the urogenital tract. Imaging facilities available included conventional radiography, contrast studies, angiography, ultrasonography and computerized axial tomography.
Information collected about each case included age, sex, specific diagnosis and short term outcome (i.e. outcome during the first admission and up to the first three months after presentation).

Results

A total number of 125 children presented with a major urogenital malformation during the study period. About 42% presented in the neonatal period while another 16% presented at ages 2-6 months (Table 1). Nearly one-quarter (23%) presented after two years of age. There was a predominance of male patients accounting for over 80% of the cases (Table 1).

Table 1. Age and sex distribution among 125 children with major urogenital malformations

		No	*%*
Age (months)			
	0 - 1 (neonate)	53	42.4
	2 - 6	20	16.0
	7 - 12	11	8.8
	13 - 24	8	6.4
	> 24	29	23.2
	Exact age unkmown	9	7.2
Sex			
	Male	106	84.8
	Female	19	15.2
Outcome			
	Discharged	106	84.8
	Died	18	14.4
	DAMA *	1	0.8

* Discharged against medical advice

The specific diagnoses are listed in Table 2. The most common were posterior urethral valves (40.0%), hypospedias (18.4%), ambiguous external genitalia (12.8%) and ectopia vesicae (11.2%). Prune-belly syndrome (Eagle-Barett syndrome) was seen in 7 (5.6%), polycystic kidneys in 5 (4.0%) and meatal stenosis/atresia in 4 (3.2%) patients.

Table 2. Diagnosis among 125 children with major urogenital malformations

Diagnosis	*No*	*%*
Posterior urethral valves	50	40.0
Hypospedias	23	18.4
Ambiguous external genitalia	16	12.8
Ectopia vesicae	14	11.2
Prune-belly syndrome	7	5.6
Polycystic kidney	5	4.0
Meatal stenosis / atresia	4	3.2
Anovestibular fistula	2	1.6
Rectovaginal fistula	1	0.8
Epispadias	1	0.8
Persistent urachus	1	0.8
Multiple urogenital - malformations	1	0.8

The age at presentation of the whole study group and of children with the four most common diagnoses is shown in Table 3.

Table 3. Age at presentation among 125 children with major urogenital malformations

Age	*All patients*	*Posterior urethral valves*	*Hypospadias*	*Ambiguous genitalia*	*Ectopia vesicae*
0 - 1	53	22	2	8	12
2 - 6	20	9	-	3	1
7 - 12	11	8	1	1	1
13 - 24	8	2	5	1	-
> 24	29	6	13	2	-
Exact age unknown	9	3	2	1	-
Total	125	50	23	16	14

Less than one-half of our cases (53/125) presented during the neonatal period. Those with hypospedias tended to present late while nearly all those with ectopia vesicae presented in the neonatal period. Of the 50 children with posterior urethral valves, 22 (44%) presented in the first month of life and 17 (34%) presented between 2 and 12 months of age. Prenatal diagnosis was made in only two cases (one case of polycystic kidneys and one of posterior urethral valves) and these were incidental findings during obstetric ultrasonography.

Overall short-term mortality was 14.4% (18/125). The 18 deaths were as follows: posterior urethral valves [11], prune-belly syndrome [3], ambiguous external genitalia [2], polycystic kidneys and meatal atresia [1]. Thus, posterior urethral valves accounted for about two-thirds (11/18) of the deaths. Mortality rate in the four most common diagnoses was 22.0 % (11/50) for posterior urethral valves, 0.0% (0/23) for hypospedias, 12.5% (2/16) for ambiguous external genitalia and 0.0% for ectopia vesicae. Among children with posterior urethral valves, those presenting in the first two months of life had a mortality rate of 45.8% (11/24), compared to no mortality (0/21) among those older than three months ($X^2 = 10.38$, df = 1, p = 0.001).

Discussion

This study described the pattern of major urogenital malformations as seen in children presenting in a major tertiary health care institution in West Africa over a ten and half years period. A retrospective study design as used in this study is probably appropriate since these malformations are individually rare. The data generated, while different from those collected from incidence and autopsy studies, but supplements other sources data.

The study has shown that posterior urethral valves is the commonest urogenital malformation seen in children in this centre. This is similar to previous findings of hospital admissions [10] and autopsy studies [7] in the same centre. Posterior urethral valves is one of the main causes of severe obstruction of the urinary tract whose cardinal clinical features as poor urinary stream and a bladder mass are easily recognised. This relative ease of recognition may account for its being most common in this series. This malformation was also associated with high mortality unlike the situation in developed countries where prenatal diagnosis and early intervention have made short-term prognosis for such patients good, with only about 3% dying during the first hospital admission [11]. The comparative seven-fold increased mortality observed in this series may be due to the fact that prenatal diagnosis of the lesions was rare, facilities for fetal surgery were not available and patients often presented with other life-threatening situations, especially infection and metabolic problems. In addition, fewer than 50% presented during the first month of life; the others presented later often with significant renal impairment. The finding of high mortality in younger patients with posterior urethral valves is probably due to the fact that early presentation is associated with more severe obstruction. Similarly, Parkhouse et al [11] have also shown that early presentation is associated with a bad long-term outcome for renal function.

The relatively low mortality with the other diagnoses (with the exception of prunebelly syndrome) is probably not surprising since these malformations are not usually lethal in themselves (e.g. hypospedias) or if so, death may occur late (e.g. ectopia vesicae). It was not possible to comment on long term outcome in these patients since the default rate was very high (irrespective of whether the child got better or worse) One factor which often determines survival and the quality of life in children with major congenital malformations in the tropics is the availability of adequate facilities for diagnosis and management. In particular, early detection using fetal ultrasonography with its sensitivity for urogenital malformations when compared to malformations of other systems [12].

While ultrasonography is increasingly used in parts of Africa, but is not yet sufficiently widespread to be used as screening tool for urogenital and other malformations. Moreover, when diagnosis is made, there may be no qualified surgeon or adequate facilities. Therefore, basic equipment and trained staff are needed to improve the management of these children. These, coupled with increased awareness of these lesions among health care providers, will probably reduce morbidity and mortality attributable to such malformations.

The main limitation of the study is the disproportionately higher frequency of the more severe malformations which is not a true picture of the actual pattern of all the urogenital malformations. However, the primary aim of the study is to identify cases that will pose challenge to the health care system and to identify possible means of early diagnosis. This study, within its limitations, has succeeded in satisfying these aims.

In summary, we have described the pattern of major urogenital malformations in children seen at the University College Hospital, Ibadan, Nigeria. The data provided supplement data from other sources and provide a framework for planning at the community level.

References

1. Mandell J, Blyth BR, Peters CA, Retik AB, Estroff JA and Benacerraf BR: Structural genitourinary defects detected in utero. Radiology 1991; 178: 193-196
2. Patten RM, Mack LA, Wang KY and Cyr DR: The fetal genitourinary tract. Radiol Clin North Am 1990; 28: 115-130
3. Callan NA, Blakemore K, Park J, Sanders RC, Jeffs RD and Gearhart JP: Fetal genitourinary anomalies: Evaluation, operative correction and follow-up. Obstet Gynecol 1990; 75: 67-74
4. Choi H, Kim KM, Koh SK, Kim KS, Woo YN, Yoon JB, Choi SK and Kim SW: A survey of externally recognizable genitourinary anomalies in Korean newborns. J Korean Med Sci 1989; 4: 13-21
5. Thein MM, Koh D, Tan KL, Lee HP, Yip YY, Tye CY and Phoon WO: Descriptive profile of birth defects among livebirths in Singapore. Teratology 1992; 46: 277-284
6. Marinho AO, Kalejaiye AO and Sobowale AO: Obvious congenital malformations in 22, 899 consecutive newborns in Ibadan. Niger J Paediatr 1987; 14: 59-65
7. Akang EEU, Osinusi KO, Pindiga HU. Okpala JU and Aghadiuno PU: Congenital malformations: A review of 672 autopsies in Ibadan, Nigeria. Pediatr Pathol 1993; 13: 659-670
8. Gutermann M, Tennstedt A. Schreiber D. Missbildungell des Urogenitalsystems im Kinderobduktiionsgut. [Malformations of the urogenital system in autopsy material of children]. Zentralbl Allg Pathol 1984; 129: 209-214
9. Jones KL. Smith's Recognizable Pattern of Human Malformation. 4 th Edition. Philadelphia: WB Saunders Company. 662
10. Adeyemo AA. Okolo CM and Omotade OO: Major congenital malformations among paediatric admissions at University College Hospital, Ibadan, Nigeria. Ann Trop Paediatr 1994; 14: 75-79
11. Parkhouse HF, Barratt TM, Dillon MJ, Duffy PG, Fay J, Ransley PG, Woodhouse CR and Williams DI: Long-term outcome of boys with posterior urethral valves. Br J Urol 1988 62: 59-62
12. Scott JE and Renwick M: Northern Region Fetal Abnormality Survey results 1987. J Pediatr Surg 1990; 25: 394-397.