Pattern of Genital Anomalies in Male Infants in Sohag Governorate

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Abstract

Background: External genital anomalies are among the most common congenital anomalies. Proper early diagnosis and management of genital anomalies are of great importance to minimize medical, psychological and social complications.

Aim: 1)explore the pattern of genital anomalies in male infants (age from the first day of birth up to two years) presented to primary health care offices in Sohag Governorate for routine vaccination, 2)determine potential environmental and parental risk factors which could be related to detected anomalies and 3) follow up male infants with cryptorchidism up the age of two years for spontaneous descent of the testis.

Patients and methods: our study included 1134 male infants who presented to primary health care offices in Sohag for routine vaccination during the period from June 2008 to March 2009. This study was conducted in Sohag city and two villages around it and also included two towns as representative of suburban area in addition to a village from each town. Male infants were examined to detect visible genital anomalies and testes were palpated along their normal anatomical pathway of descent and cases of cryptorchidism followed at three months interval up to two years to detect spontaneous descent of the testis .A clinical data sheet was filled for all male participants including demographic data, parental data and results of clinical examination.

Results: There were 129(11.3%) cases with genital anomalies among the screened 1134 male infants ,Various abnormalities in the form of ,hydrocele , cryptorchidism, hypospadias, indirect inguinal hernia, phimosis and epispadias were diagnosed. No spontaneous descent of the testis was detected in cases with cryptorchdism during the follow up period. Family history of genital anomalies, residency in rural area, multiparity and cesarean section were detected as risk factor associated with genital anomalies .

Conclusion: This study in Sohag Governorate, Upper Egypt showed a relatively high prevalence of genital anomalies. Therefore we recommended more studies including larger population sizes to detect the actual data about genital anomalies and associated risk factors. Increasing clinical education of physicians about this problem, as well as raising the awareness of nurses and parents is important.

Introduction

External genital anomalies are among the most common congenital anomalies. Proper early diagnosis and management of genital abnormalities are of great importance to minimize medical, psychological and social complications.1

A study of genital anomalies in male pediatrics has become a matter of interest nowadays, because of the effect of these anomalies on the future of life & fertility. Most data about these patterns are almost exclusively derived from hospital-based birth defect registers which are sensitive to selection bias and incomplete reporting.2 Genital anomalies of male children include those affecting shape of the penis, urethral opening (meatal

stenosis, epispadias and hypospadias) and scrotum (bifid scrotum, cryptorchidism and hydrocele). Also, they include ambiguous external genitalia and stenosed prepuce (phimosis).3 Among all anomalies, cryptorchidism and hypospadias are the most frequent congenital anomalies detected in early childhood.4 The prevalence of cryptorchidism has increased over the last few decades in several countries. The aetiology is multifactorial and includes hormonal and molecular factors. Many authors have attributed this increase in the rates of cryptorchidism to estrogenic effect of environmental endocrine disruptors. This estrogenic effect may be derived from maternal or paternal occupation or life style.5 It has been speculated that spontaneous testicular descent occurs in more than 70% of newborns with cryptorchidism. Diagnosis of cryptorchidism depends on clinical examination by detection of empty scrotum. However, laparoscopy remains the single most accurate modality for diagnosis and localization of impalpable and ascended gonads.6 It seems essential that cases of cryptorchidism are diagnosed early and treated early because if a testis is left in its undescended or maldescended position for a prolonged period, there are risks of atrophy, torsion, malignant changes and subfertility.7 Hpospadias is a midline fusion defect of the male ventral urethra is a relatively common genital anomaly. The anatomical location of the misplaced urethral meatus determines the severity of this anomaly with severity increasing from distal to proximal.8 The most common forms of hypospadias (glandular and penile) often appear as an isolated anomaly whereas about 20% are classified as scrotal and perineal types. These latter forms frequently occur in association with other genital anomalies such as microphallus, bifid scrotum and cryptorchidism.9 Little is known about environmental risk factors for genital anomalies but many studies focused on potential endocrine disruptors in parental diet, occupation, pregnancy aspects, life style and personal characteristics.10

Aim of the work:

This study was done to detect pattern of genital anomalies in male infants presented to primary health care offices in Sohag Governorate for routine vaccination and to determine potential environmental and parental risk factors that could be related to detected anomalies.

Patients and Methods:

This study and the study protocol were approved by the local postgraduate institutional research committee in Sohag Faculty of Medicine.

The study was conducted in Sohag city and two villages around it as representative of urban and rural areas, respectively. In Sohag city we checked male infants attending the second health office(n=133) & Salama Abdallah health centre (n=128) (in the Western area), the fourth office (n=190) (in the Eastern area) and El Mazaloa health office (n=64) (in the Southern area) and Gizerat shandawel (n=181) (in the North) ,also this study included two towns Tema (n=92) (to the North of Sohag) and EL Baliana (n=131) (to the South of Sohag) as representative of suburban areas. In addition, the study included Meshta (n=155) (a village from Tema) and El Eslah (n=60) (a village from El Baliana). A clinical data sheet was filled for all male infants participant.

Data were collected from all subjects and the data included:

Gestational data: included age of mother during pregnancy, diet, drug intake, radiation exposure and any medical diseases, fever, problems during pregnancy or TORCH , previous abortion and hormonal contraception use before pregnancy ,presentation of the infant and type of labour, previous babies with genital anomalies.

Birth & infantile data included age of birth, birth weight, birth order and other data included age ,weight, height and head circumference at time of examination of the infant.

Socio-demographic and environmental data for parents: included name, age, history of consanguinity, occupation, education and place of residency, special habits, medical diseases, medication, radiation exposure, family history of genital anomalies and history of infertility.

Clinical assessment of the patients:

Medical and family histories were obtained from all patients and complete genital examination was performed to detect any visible anomalies such as meatal opening, prepuce, penis and scrotum.

Testicular palpation took place in anxiety free and warm place, testicular position was described after manipulation of the testis to the most distal position along the pathway of normal descent, using firm but not forced traction. The testis was considered cryptorchidism if found in a high scrotal, suprascrotal, inguinal position or was non palpable. Retractile testis was considered to be a normal variant.11 Each boy with cryptorchidism was re-examined at 3 months interval and up to 2 years to detect spontaneous descent . General examination was done to detect associated anomalies with genital anomalies.

Statistical analysis:

All collected data were recorded in excel data sheet. Data entry and statistical analysis were performed using the statistical package for the social science (SPSS version 10 program). Qualitative variables were presented as frequencies and percentages and compared using Chi-square test. Quantitative variables were presented as mean ± standard deviation (SD) and were compared by independent student t- test and logistic regression analysis was done to detect associated risk factors for male genital anomalies. P-value ‹ 0.05 was considered significant.

Results

Our study included 1134 male infants, 129(11.3%) had genital anomalies, scrotal swelling were detected in 107 male infants and 7 infants had underdeveloped scrotum, also cryptorchidism was detected in 8 infants (6.2% of the anomaly) , one of them had bilateral presentation and 7 infants had unilateral presentation (4 cases were on the right side and 3 cases on the left side). Hydrocele was found in 107 male infants (82.9% of the anomaly), 91 of them were with bilateral presentation and 16 cases were on the right side. According to penile and urethral anomalies, 8 male infants (6.2% of the anomaly) had hypospadias, 6 of them the urethral meatus was found in subcoronal position (2 of them with mild chordee) and 2 cases had glandular hypospadias. One patient (0.7%) had phimosis. Indirect inguinal hernias were detected in 4(3.1%) male infants (3 cases on the right side and one with bilateral presentation) and one patient (0.7%) had glandular epispadias. Follow up of the patients of cryptorchidism every 3 months was done for the eight diagnosed cases, 7

of them had palpable UDT (6 at inguinal level and one suprascrotal) and one had impalpable testis, two cases were missed during the follow up, the reminder 6 cases ,no spontaneous descend was detected then orchidopexy was done for 5 cases after ultrasonography at different age ranging from 9 months up to 2 years and one case was treated by hormonal therapy at age of 6 months (Table 1).

Table 1: The distribution of male infants genital anomalies.

Type_of_anomlies (total number= 129)	
	Scrotal
Swellings	**107**
Underdeveloped	**7**
	Testicular
Cryptorchidism:-	**8**
Cryptorchidism(unilateral)	**7(4 RT – 3LT)**
Cryptorchidism (bilateral)	**1**
Hydrocele:-	**107**
Hydrocele (unilateral)	**16 (RT)**
Hydrocele (bilateral)	**91**
	Penile and urethral
Epispidus	**1 (glandular)**
Hypospidus	**8 (6 subcoronal (2with mild chordee) – 2 glandular)**
Phimosis	**1**
	Others
Inguinal hernia	**4 (3 RT – 1 BIL)**

Male infants genital anomalies were significantly associated with residence in rural area (12.4%) (p-value=0.04), cesarean section(16.5%) (p-value =0.03), family history of genital anomalies(50%) (p-value=0.01), X-ray exposure during pregnancy (100%) (p-value =0.05), multiparity(12.2%) (p-value =0.05) and farmers(18%) (p-value=0.04). Also anomalies were higher in cases with history of consanguinity (12.2%) and previous baby with genital anomalies (17.9%) but without statistical significance. Table(2)

Table(2):the comparison between male infant with genital anomalies according to place of residence ,type of labour ,history of consanguinity, family history, previous baby with genital anomalies parity and occupation of father versus those without anomalies.

Variables	**Genital anomalies**		**p-value**
Residence	**Present %**	**Absent %**	
Rural(683)	85(12.4%)	598(87.6%)	**0.04**
urban (451)	44(9.8%)	407(90.2%)	
Type of labour			
Normal(850)	82 (9.6%)	768 (90.4%)	**0.03**
C.s(284)	47 (16.5%)	237 (83.5%)	
History of consanguinity			
Yes(394)	48 (12.2%)	346 (87.8%)	**0.53**
No(740)	81 (10.9%)	661 (89.1%)	
Family_history of genital anomaly			
Yes(4)	2 (50.0%)	2 (50.0%)	**0.015**
No(1130)	127 (11.2%)	1003 (88.8%)	
Previous-baby With genital anomalies			
No(1106)	124 (11.2%)	982 (88.8%)	**0.3**

Yes(28)	5 (17.9%)	23 (82.1%)	
X-ray exposure during pregnancy			
Yes(1)	1 (100.0%)	0 (.0%)	**0.05**
No(1133)	128 (11.3%)	1005 (88.7%)	
Parity			
Primi(335)	31 (9.2%)	304 (91.8%)	**0.05**
Multi(799)	98 (12.3%)	701 (87.7%)	
Occupation			
Farmer(111)	20(18%)	91)(82%	**0.04**
Not farmer(1023)	109 (10.4%)	914 (89.6%)	

Logistic regression analysis of the significantly different variables was used. The most causal associated risk factors with genital anomalies were family history, C.S, multiparity and residence in rural area. Table(3)

Table3: the most causal risk factors for male infants genital anomalies

	Logistic regression			
	B	**OR**	**Sig.**	**Exp(B)**
Mother_Family_history	**2.093**	**4.193**	**0.02**	**0.823**
C.S labour	**0.392**	**3.499**	**0.03**	**0.676**
Parity multi	**0.224**	**2.066**	**0.03**	**0.666**
Rural	**0.534**	**1.129**	**0.04**	**0.586**
Constant	**3.194**	**16.885**	**0.000**	**24.381**

Discussion

External genital abnormalities represent the most common congenital anomalies. Proper not delayed diagnosis and treatment of certain abnormalities (undescended testis and hypospadias) is of great importance for future fertility potential of children.12

Malformations such as (hypospadias, epididymal anomlies and inguinal hernia) are often observed in cases of cryptorchidism and this may direct the search for potential similar risk factors.13

In our study, 1134 male infants attending the primary health care offices in Sohag for routine vaccination were examined. 129 (11.3 %) male infants had genital anomalies. Communicating hydrocele constitutes the most common genital anomaly (107 cases (82.9 %) of detected anomalies and 9.4% of all examined infants) and this is in agreement with the concept of patent processus vaginalis which is found in 80% - 90% of full term male infants, Our results agree with those of Mazen et al.1 The second most common genital anomaly was undescended testis which was affecting 8 patients (6.2 % of the genital anomalies) and (0.7 % of the all screened male infants), This is similar to that of Scorer14 who reported the prevalence rate of cryptorchidism 0.8% at the age of one year along with Docimo 15 who reported that up to one third of premature male newborns are born

with an undescended testicle and 3 to 5% of term male infants are affected by three months of age, the incidence is reduced to 0.8% between three months of age and adulthood the incidence does not change. Eight patients had hypospadias (6.2% of genital anomalies and 0.7% of examined male infants), this is similar to that reported by Paulozzi 16 who said that hypospadias is one of the most common birth defects in the United States, affecting as many as 1 in 125 newborn male and also that is in agreement with Gallentine et al17 and Baskin18 who found that hypospadias has been estimated to be 0.4 to 8.2 per 1000 live male births. Four patients had indirect inguinal hernia (0.3% of all examined infants and 3.1% of genital anomalies) and this is in agreement with Lee et al19 who found that the incidence of inguinal hernia and hydrocele varies from 0.4 to 13.4%. In the present study, One patient had epispadias (0.08% all examined infants and 0.7% of male genital anomalies) and it was of glandular type. Dees20 said that epispadias occurs more often in combination with bladder extrophy (1 in 40.000 births) than as an isolated event (1 in 90.000), also Anne-Karoline Ebert et al21 who found that epispadias estimated in an average rate of 2.4 per 100.000 births. And also, one patient had phimosis, Osta22 who said that by 3 years of age, 90% of foreskin can be retracted and lysis of the natural adhesion between the glans and prepuce occurs as the penis grows and smegma accumulates. In our study 30.8 % of all examined male infants were circumcised

Fernandez et al23 found that urogenital malformations were common in infants living in rural area (32 %) than those living in urban area (29%) and this is similar to our result as male infants genital anomalies were common in rural area(12.4%) than urban area(9.8%).

In the present study, there was a significant association between genital malformations and delivery by cesarean section (16.5 %) versus normal delivery (9.6%) and P-value = 0.03 and this is similar to that reported by Aschim et al24, Fernandez et al23 and Damgaard et al25. Cesarean section reflects risk factors such as (pre maturity and abnormal presentation), but itself in not an important risk factor. In our study, there was no statistically significant difference between the mean age of mother during pregnancy of male infants with genital anomalies (27.1 ± 5.6) years versus those without anomalies (27.2 ± 5.2) years (p-value = 0.7), also there was no statistically significant difference between the mean age of father of male infants with genital anomalies (35.2 ± 6.5) years versus those without anomalies (34.8 ± 6.7) (p - value = 0.5), so no effect of parental age on urogenital malformations was observed.

Our study showed that parental consanguinity was an important factor for the development of male infants genital anomalies. There was a statistical significant association between male infants genital anomalies and family history of genital anomalies (p-value = 0.01) and this is agreement with Bauer et al26 and Jones and Young27 and Weidner et al 28 who reported that hypospadias and genital malformation have been found to have an increased risk with familial incidence. Male infants genital anomalies were more common in patients with brothers having genital anomalies (17.9%) than those without (11.2%) and this is similar to that reported by Weidner et al28 and these results support the role of genetic

hypothesis in the aetiology of genital anomalies.

In the present study, there was a statistically significant association between the development of genital malformation and material X ray exposure during pregnancy (p - value = 0.05) as Sweet and Kinzie29 reported that in utero exposure to radition, increase the risk for malformation and subsequent increase likehood of development of neoplasia. Male genital anomalies were more common in multipara (12.3%) than primipara (9.2 %) and that was statistically significant (P- value = 0.05) but this is not as Swerdlow et al30 found the risk of cryptorchidism was higher in boys born to primipara .

There was no statistically significant association between male infant genital anomalies and smoking status as genital anomalies were more or less equal in smoker(11.2%) and non smoker fathers(11.4%) and p-value=0.7 and this is similar to Fernandez et al23 who reported no association between cryptorchidism and or hypospadias and smoking habit.

Paternal occupation as farmer was significantly associated with male infants genital anomalies and p-value=0.04 ,this is in agreement with Rueda-Domingo et al31 who reported cryptorchidism was related to father employment in agriculture..

In the present study, the most causal risk factors were family history then C.S, then multiparity and residency in rural area.

Conclusion:

In conclusion, our results show increase in the prevalence of genital anomalies, so we recommended accurate and early diagnosis of congenital malformation which is the key to proper management of cases. Premarital counseling is advised, especially in the presence of parental consanguinity and family history of anomaly.

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