

Prevalence of genitalia malformation in Iranian children: findings of a nationwide screening survey at school entry

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Abstract

Background: Ambiguous genitalia is a hereditary disorder that usually requires early attention and detection. The discovery of ambiguous genitalia in a neonate is a situation that could be difficult to manage, not only because of complications such as salt-losing, but also due to the importance of sex determination before psychological gender could be established. Awareness of the prevalence of ambiguous genitalia can affect the attitude and consideration of physicians and related medical personnel about disease in different communities. So in this study, the prevalence of ambiguous genitalia and undescended testes (UDT) in Iran was reported.

Materials and Methods: This national study was conducted in 2009-2010 as part of the routine screening examinations at school entry in Iran. The physical examinations were performed for students at entry to three school levels by physicians and medical personnel. Execution and conduction of this program was the duty of the University of Medical Sciences in each province.

Results: On average, the prevalence of ambiguous genitalia was 0.04% at national level (0.03%, 0.05%, and 0.03% at 6, 12, and 15 year olds, respectively). The prevalence of ambiguous genitalia was not significantly different according to age group and living area. The average of UDT prevalence in the whole country was 0.13%. The prevalence of UDT was higher at elementary school level than in the other two levels.

Conclusion: Although the prevalence of genitalia abnormalities was not high in the school students in Iran, given the importance of the issue and in order to find the ambiguous genitalia or UDT, medical examinations and parental notification should be taken seriously at an earlier age.

Key Words: Ambiguous genitalia, screening examinations, undescended testes

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Received: 22.03.2013, **Accepted:** 25.08.2013

Access this article online	
Quick Response Code:	Website: www.advbiores.net
	DOI: 10.4103/2277-9175.125648

INTRODUCTION

The birth of a newborn is one of the most dramatic events in a family unit, and the initial question is generally “is it a girl or a boy?”. The newborn infant with ambiguous external genitalia often comes as a shock to the parents as well as the physicians.^[1] In the majority of infants at birth, sex assignment is direct.^[2]

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How to cite this article: Armanian AM, Kelishadi R, Ardalan G, Taslimi M, Taheri M, Motlagh M. Prevalence of genitalia malformation in Iranian children: Findings of a nationwide screening survey at school entry. *Adv Biomed Res* 2014;3:36

Ambiguous genitalia is a congenital disorder that usually requires early attention and detection. The finding of ambiguous genitalia in a neonate is a situation that could be difficult to manage, not only because of complications such as salt-losing, but also due to the importance of sex determination before psychological gender could be established.^[3,4] Physical examination, particularly chary palpation to find gonads at the genital folds or in the inguinal region is the most important way to detect.

Furthermore, hormonal, genetic, radiographic, and molecular scrutiny is required to find out the etiology.^[3,5]

Presentations of ambiguous genitalia may include “bilateral cryptorchidism, perineal hypospadias, clitoromegaly, posterior labial fusion, phenotypic female appearance with a palpable gonad (with or without inguinal hernia), hypospadias, and unilateral no palpable gonad”.^[6]

Given the importance of the issue, awareness of the prevalence of ambiguous genitalia can affect the attitude and attention of physicians and related medical personnel about disease in different communities. There are inadequate data on the prevalence of ambiguous genitalia;^[1] it is expected that the overall frequency of ambiguous genitalia is one in 5500.^[7,8] The etiologies of almost 50% of all cases of ambiguous genitalia in the neonatal period are congenital adrenal hyperplasias and mixed gonadal dysgenesis.^[9] Abnormalities, which could be quickly assessed, were observed in approximately one in 4500 live births, in another study.^[10]

It is possible that the ambiguous genitalia are not found at birth examinations, for any reason; or may be denied by parents to prevent emotional problems for their child. Therefore careful evaluation and consideration seems to be necessary in another proper setup as office-based physical examination or screening through school-based screening programs.

Thus, examination of the genitalia is considered as part of the school entry screening programs in Iran. The current nationwide study aimed to determine the prevalence of ambiguous genitalia among school students.

MATERIALS AND METHODS

This nationwide study was conducted in 2009-2010 as part of the routine screening examinations at school entry in Iran. This screening program is conducted every year by the Iranian Ministry of Health and

Medical Education and the Ministry of Education and Training. Implementation of this program was the responsibility of the University of Medical Sciences in each province.

The examinations were performed for all students at entry to three school levels, that is, at the age of 6 years (at entry to elementary school), 12 years (at entry to middle school), and 15 years (at entry to high school). The examination consisted of two parts: preliminary assessment by school health care providers and then medical examinations by physicians.

Some diseases or defects was evaluated with this method such as gum disease and dental caries, endocrine disorders, heart and vascular disease, abdominal disease, growth disorders, behavioral disorders, ambiguous genitalia, undescended testes (UDT), vision and hearing problems, spinal deviation and etc. This examination is performed in Iran at the various school levels by physicians and medical personnel. Among the diseases that had great importance in the national screening program were ambiguous genitalia and UDT. Therefore, trained health personnel for symptoms of genital ambiguity started the physical examinations, and patients were referred to specialists.

Here, we report the results of detection of ambiguous genitalia and UDT (for boys) in routine examination (screen) for ambiguous genitalia (for all students at entry to three school levels).

In this study, statistical analysis was performed by descriptive statistics. In this regard, the prevalence of diseases was evaluated in provincial and national levels.

RESULTS

The medical screening examinations had a coverage of 84.4% in the whole country, consisting of 94.7% in the elementary schools, 85.2% in the middle school, and 74.5% in the high school students [Figure 1].

On average, the prevalence of ambiguous genitalia was 0.04% at the national level. The prevalence of ambiguous genitalia was 0.03%, 0.05%, and 0.03% at entry to elementary school, at entry to middle school, and at entry to high school, respectively. Therefore, the prevalence of ambiguous genitalia was not significantly different according to age group. Also the prevalence of ambiguous genitalia was similar according to living area, that is, urban versus rural (0.04% vs. 0.03%, respectively). Nevertheless, responsible universities for screening program in each province reported different results with regard to prevalence.

University of Kashan, East Azarbaijan, and North Khorasan reported the highest rate of genital ambiguity (0.22%, 0.18%, and 0.16%, respectively). This prevalence was also higher than national average in University of South Khorasan, Jahrom, Markazi, and Razavi Khorasan [Figure 2].

The average percentage of UDT prevalence in the whole country was 0.13%. The prevalence of UDT at Qom University, North Khorasan, Razavi Khorasan, Jahrom, Hamedan, Kurdistan, South Khorasan, Shiraz, West Azerbaijan, Golestan, Hormozgan, and Markazi had a rate higher than the national average

[Figure 3]. The prevalence of UDT was higher at the elementary school level than in the other two levels (0.26% vs. 0.07% and 0.02%).

DISCUSSION

Ambiguous genitalia and UDT should be diagnosed in the neonatal period; however, we found cases with undiagnosed disorders till school age. This highlights the importance of accurate physical examination at birth and in further medical visits of children. Clinicians and other professionals should diagnose the disorder whenever there is an opportunity.

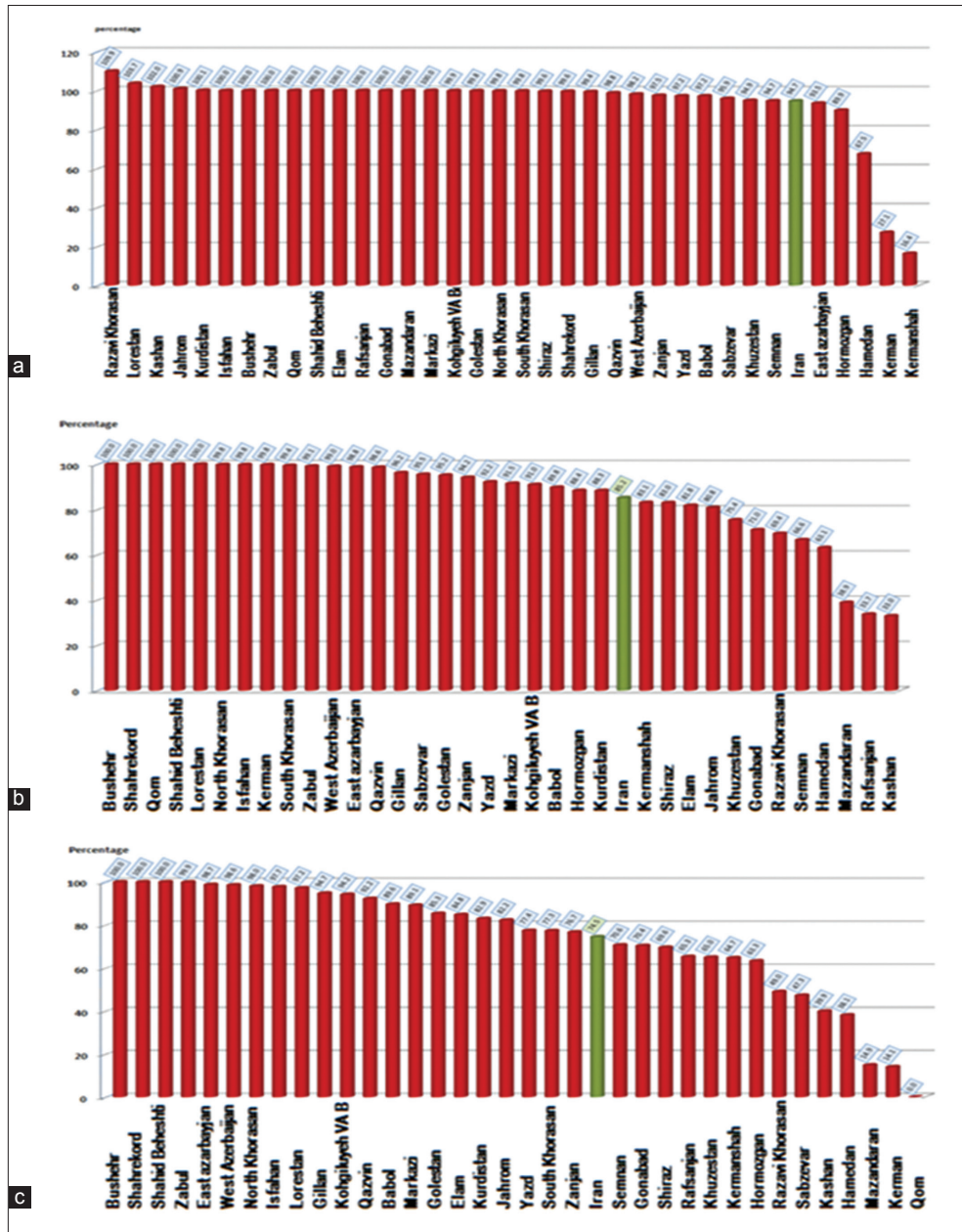


Figure 1: Frequency of examined students in various parts of Iran in 2009-2010. (a) At entry to primary school; (b) at entry to middle school; (c) at entry to high school

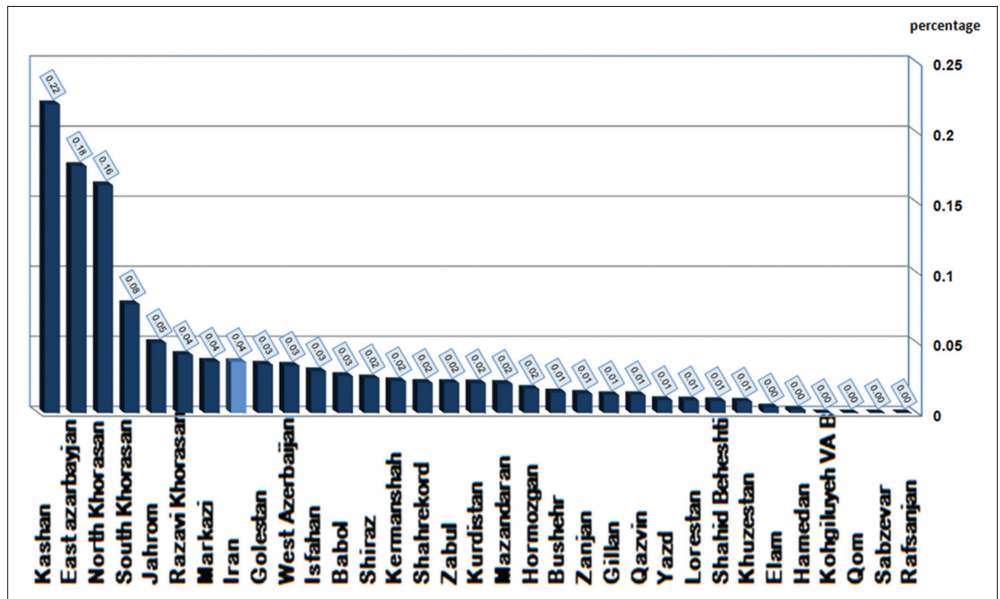


Figure 2: Comparison of the frequency of ambiguous genitalia in students screened in different cities in Iran

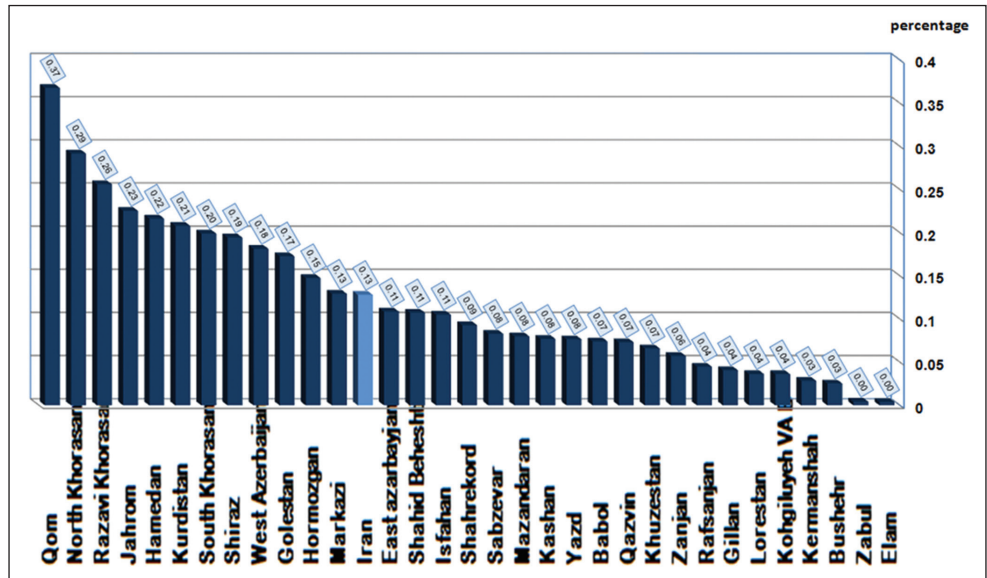


Figure 3: Comparison of the frequency of undescended testes in students screened in different cities in Iran

The prevalence of ambiguous genitalia was 1/5500 live births in the study by Sax *et al.*^[8] Although adequate data is not available on the prevalence of ambiguous genitalia, in some studies such as Hughes’s study in Philadelphia, the disease occurs in approximately one in 4500 live births^[10] and in Karbasi *et al.*’s, study the prevalence of ambiguous genitalia and UDT *at birth* was 3.18% and 1.36%, respectively, in Yazd, Iran.^[11] In our study the overall prevalence of ambiguous genitalia at school ages in the whole country was 0.04% and prevalence of ambiguous genitalia and UDT *at school ages* was 0.01% and 0.08%, respectively, in Yazd, Iran.

Hack *et al.*, found in their study that the occurrence of UDT for 6 year, 9 year, and 13 year olds was 1.2, 2.2, and 1.1%, respectively, in the Netherlands.^[12] Sijstermans *et al* concluded that “At birth, in term and/or birth weight >2.5 kg infants, the UDT rate ranged from 1.0% to 4.6%, and in premature and/or birth weight <2.5 kg infants from 1.1% to 45.3%. At the age of 1 year, UDT in term and/or birth weight >2.5 kg infants was seen in 1.0%-1.5%, at 6 years in 0.0-2.6%, at 11 years in 0.0-6.6%, and at 15 years in 1.6-2.2% of boys” in a systematic review.^[13] In our study the overall prevalence of UDT in the whole country was 0.13% (0.00-0.37%). We found that occurrence

of UDT for 6 year, 12 year, and 15 year olds was 0.26, 0.07, and 0.02%, respectively, in the in whole country. The difference in our results with some other studies may be due to differences in the accuracy of the examination. Therefore, higher prevalence in other studies may indicate the need for more precision in our medical examinations.

Yegane *et al* studied 3205 school boys. They detected retractile testes in 1.22% boys, UDT in 1.12% boys, and hydrocele in 0.87% boys, but no ambiguous genitalia were seen in that study in the west of Iran. The interesting thing was that 60.1% of the parents were not conscious of their male children's abnormalities.^[14]

Ozoemena and Mbah *et al* recorded 6.8% of congenital abnormalities of external genitalia (a high prevalence rate) on school age students, which were undiscovered from birth in a study in Nigeria.^[15]

Therefore, to find the ambiguous genitalia or UDT, medical examinations and informing parents should be considered seriously.

It is noteworthy to mention that although all health professionals received similar training for doing the physical examinations in this screening survey, some differences in the prevalence rates of abnormalities reported in various cities might be because of the differences in the accuracy of physical examination by clinicians with different expertise levels.

CONCLUSION

Although the prevalence of genitalia abnormalities was not high in the school students in the whole country, such disorders should be diagnosed in early life and even the low prevalence of abnormalities documented in this study are of crucial importance. Therefore, given the importance of issue and in order to find the ambiguous genitalia or UDT, medical examinations

and parental notification should be taken seriously at an earlier age.

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Source of Support: Data of a national screening program were used in this study. **Conflict of Interest:** None declared.