SCREENING PROGRAM FOR URO-GENITAL CONGENITAL ANOMALIES IN CHILDREN

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Abstract

Aims. *The aim of the study is the evaluation of the results of a screening program for the diagnosis of uro-genital congenital anomalies in a school-age population of children.*

Patients and methods. *Between 2005 and 2008 we initiated a screening program for uro-genital anomalies and we examined the children from 10 schools in Cluj, Sibiu and Mureş Counties. The evaluation included clinical examination and ultrasound evaluation of the kidneys and the urinary tract. The following parameters were assessed: presence, location and symmetry of size of the kidneys, the presence of hydronephrosis, dilatation of the ureters, post-void residual urine, and presence of the testicles in the scrotum, existence of phimosis and location of the urethral meatus.*

Results. *The screening of 1006 children (aged 5-18 years) yielded 93 (9.3%) patients with uro-genital anomalies. There were eight children diagnosed as having unilateral cryptorchism, eight unstable testes, 31 cases of phimosis or hypertrophy of the frenulum, eight hypospadias, five hydrocele, three children with bilateral cryptorchism and one case each with varicocele, micropenis and sperm cord cyst. The ultrasound evaluation revealed 14 duplex renal units without hydronephrosis, six cases of hydronephrosis, three cases of renal cysts, two cases of renal ectopia and two cases of unilateral renal agenesia. Surgical intervention was planned in 43 cases (4.3%).*

Conclusions. *Screening for uro-genital congenital anomalies using clinical examination and ultrasound is effective. The higher incidence of the genital abnormalities should draw the attention of the pediatricians upon the genitals during routine examinations.*

Keywords: early diagnosis, screening, urogenital anomalies.

PROGRAM DE SCREENING PENTRU ANOMALIILE URO-GENITALE CONGENITALE LA COPII

Rezumat

Introducere. *Ne propunem să evaluăm rezultatele obţinute printr-un program de screening pentru depistarea anomaliilor uro-genitale la copii, realizat într-o populaţie pediatrică de vârstă şcolară.*

Material şi metodă. *În intervalul 2005-2008 am realizat un screening pentru anomalii uro-genitale la copii. Programul a constat în examinarea copiilor de vârstă şcolară, de la 10 şcoli din judeţele Cluj, Sibiu şi Mureş. Evaluarea a cuprins examenul clinic şi ecografia aparatului urinar. Au fost înregistraţi următorii parametri: prezenţa, localizarea şi simetria de dimensiune a rinichilor, prezenţa hidronefrozei, dilataţia ureterelor în segmentul retrovezical, reziduul vezical postmicţional, prezenţa testiculelor în scrot, fimoza şi localizarea meatului uretral extern.*

Rezultate. *Am examinat 1006 copii, cu vârste între 5 şi 18 ani şi am constatat*

existenţa unor afecţiuni uro-genitale de gravitate variabilă la un număr de 93 copii (9,3%). În cazul băieţilor au fost diagnosticate opt cazuri de criptorhidie unilaterală, opt cazuri de testicule instabile, 31 cazuri de fimoză sau hipertrofie a frenului prepuţial, opt cazuri de hipospadias, cinci copii cu hidrocel, trei copii cu criptorhidie bilaterală şi câte un caz de varicocel, chist de cordon spermatic şi micropenis. În urma examenului ecografic am identificat 14 pacienţi cu duplicaţie pielo-ureterală fără hidronefroză, şase copii cu hidronefroză, trei copii cu chiste renale, două cazuri de ectopie renală, două cazuri de rinichi unic congenital. S-a stabilit indicaţie operatorie la un număr de 43 de pacienţi (4,3%).

Concluzii. *Având în vedere numărul mare de anomalii congenitale depistate la copii aparent sănătoşi, programul de screening este eficient şi necesar. Incidenţa mare a afecţiunilor genitale trebuie să atragă atenţia medicilor de familie şi medicilor pediatri asupra acestor organe în cursul examinărilor de rutină pe care le efectuează.*

Cuvinte cheie: depistare timpurie, screening, anomalii urogenitale.

INTRODUCTION

Approximately 10% of children are born with congenital uro-genital anomalies. For a long period of time they may have only non-specific symptoms (recurrent urinary tract infections, fever, failure to thrive and enuresis). In many cases the diagnosis is made after a considerable lapse of time, sometimes when chronic renal failure is already installed [1]. The congenital urinary anomalies are the main cause for chronic renal failure in children [2,3]. They generate both a material burden for the society and family dramas that might be avoided by an early diagnosis. We believe that any effort is worthwhile in order to reduce the negative impact of these anomalies.

If the urinary congenital anomalies are not diagnosed and treated in time, the child is at risk for chronic renal failure or other complications. The occurrence of such diseases has a decisive impact on life expectancy, quality of life and financial costs. Early diagnosis and treatment may provide a quality of life similar to that of healthy children with a lower financial burden for society. In a similar way, untreated genital anomalies may lead to primary infertility, carcinogenesis and psychological problems (inferiority complexes) [4].

International public health policies are currently oriented towards prevention and early diagnosis of uro-genital anomalies. The main priority is the attempt to minimize their negative effects, by means of an early diagnosis of affected children, before irreversible complications occur. This can be carried out on two levels:

- prenatal ultrasound screening for urinary anomalies;
- screening of all the apparently healthy children [5].

Manuscript received: 22.09.2011
Received in revised form: 30.11.2011
Accepted: 07.01.2012
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The prenatal ultrasound examination and the postnatal clinical examination immediately after birth can discover most of the uro-genital anomalies. There are still a number of anomalies that are skipped by this protocol: the testicular descent can be completed several months after birth (especially in pre-term boys), the foreskin may be fully retracted only at the age of 3 and several children might have never been examined by ultrasound for a variety of reasons. The need is there for a diagnostic "filter" that should include all the children, several years after birth.

There are a small number of published studies regarding screening programs either for urinary or genital anomalies in several countries in the European Union, Japan and the United States of America [6].

The aim of this study is the evaluation of the results of a screening program for the diagnosis of uro-genital congenital anomalies in a school-age population of children.

MATERIAL AND METHOD

Between 2005 and 2008 we realized a screening program for the diagnosis of uro-genital anomalies in children. We examined school-age children in 10 schools in Cluj, Sibiu and Mureş Counties. We were helped by the school physicians and the school managers in organizing the examinations. We used in this respect the school's medical facilities or a properly equipped classroom (figure 1) and we scheduled the children for examination (usually one class per hour).

We used a portable Medison SA 600 ultrasound machine with a 3.5 MHz transducer.

The screening program was financed by a CEEX grant (154/2006: "Screening, prophylactics and correction of congenital uro-genital malformations in children, in the era of minimally invasive treatment techniques (laparoscopy, endoscopy)" – SCANURGENT).

Figure 1. Examining a child in a primary school classroom in Chinteni, Cluj County.

The investigation protocol consisted of:

• medical history, in order to discover typical symptoms (fever, recurrent urinary tract infections, sphincter control etc);

• clinical examination of the external genitalia for genital anomalies (phimosis, hypospadias, cryptorchism);

• ultrasound examination of the urinary tract.

We recorded the following parameters:

• position of the urethral meatus;

• absence of one or both testicles from the scrotum;

• phimosis (a disproportion between the size of the gland and the opening of the foreskin).

During the ultrasound examination we recorded:

• for the kidneys: presence, size symmetry, position, abnormal ecogenity, dilation of the renal pelvis;

• for the ureters: dilation of the terminal segment;

• for the urinary bladder: anomalies of the shape and the presence of post void residual urine.

Clinical or ultrasound suspicion of an uro-genital anomaly prompted us to address the child to a Pediatrics Clinic for a detailed diagnostic evaluation. For those children that required surgery, we offered the possibility of being treated in the Clinical Urology Department of the Clinical Municipal Hospital in Cluj-Napoca. The medical treatment and further surveillance were done by the family doctors or the pediatricians.

RESULTS

We examined 1006 children (between 5-18 years of age) and we have found uro-genital anomalies in 93 patients (9.3%). The 497 boys in our study group had the anomalies presented in table no. I.

Table I. Genital congenital malformations.

Type of congenital genital malformation	Number of cases	% (N=497)	
unilateral cryptorchidia	8	1,6%	figure 2
unstable testes	8	1,6%	figure 3
bilateral cryptorchidia	3	0,6%	
phimosis or hipertrophy of the frenulum	31	6,2%	figure 4
hypospadias	8	1,6%	figure 5
hydrocele	5	1%	
varicocele	1	0,2%	
micropenis	1	0,2%	
spermatic cord cyst	1	0,2%	
Total	**66**	**13,3%**	

The ultrasound evaluation of all the 1006 children revealed the anomalies presented in table no. II.

Table II. Urinary tract congenital anomalies.

Urinary tract congenital anomalies	Number of cases	% (N=1006)
duplex renal unit without hydronephrosis	14	1,4%
hydronephrosis	6	0,6%
simple renal cysts	3	0,3%
renal ectopia	2	0,2%
unilateral renal agenesia	2	0,2%
Total	**27**	**2,7%**

DISCUSSIONS

This is to our knowledge the first combined screening program for urinary tract and genital anomalies in Romania. The large numbers of boys that were identified carrying genital malformations (13.3%) emphasizes the need for this kind of programs. The idea of a combined screening program, both for urinary tract and genital tract anomalies, was received very well by the urologists both in Romania and abroad; we have presented our experience at prestigious international scientific meetings [5,7].

Cryptorchidia is the most frequent genital congenital anomaly in boys, occurring in 1% of one-year-old children; it is defined by the absence of one or both testicles from the scrotum. These may be situated anywhere on the normal descending tract (cryptorchism) or they may have descended from the abdominal cavity, but on an abnormal tract (testicular ectopy).

Clinical examination includes the inspection of the scrotal region, the perineum, the inguinal area and the upper thigh. This is followed by the palpation of the inguinal area, the examinator "milking" the inguinal canal and pushing the gonad towards the scrotum. Ultrasound examination and computed tomography are usually performed in cases when the testicle is not accessible by clinical examination, but their sensibility is lower than that of the diagnostic laparoscopy. The diagnostic method that can establish with the highest degree of certainty the patient's gonadic status (allowing also initiation of treatment) is diagnostic laparoscopy. An absent testicle and the hypertrophy of the contralateral one may suggest unilateral testicular agenesis,

but the low specificity of this association cannot eliminate diagnostic surgical intervention.

In our series of male patients the prevalence of cryptorchidia was 2.2%. The patient with undescended testis runs the risk of infertility. To prevent it corrective intervention is recommended during the first two years of life, this period of time offering the highest chance of a complete recovery of the testicular function. This is why we recommend systematic evaluation of the presence of testicles in the scrotum for all newborn babies. For those with one or both testicles missing, their descent must be expected until no later than one year of age. Another risk associated with cryptorchism is the occurrence of a malignant tumor in the non-descended testicle, 10-15 times higher in the case of patients with cryptorchism. Recent studies suggest that the surgical intervention (orchidopexy) lowers the risk of malignancy (OR 5.4 without surgery and 2.23 after surgery) [8,9].

Figure 2. Left unilateral cryptorchism.

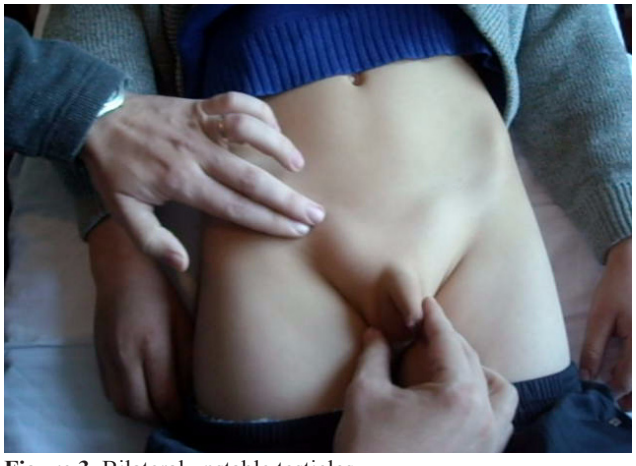

Figure 3. Bilateral unstable testicles.

Phimosis defines the situation in which the dimension of the glans is larger than that of the preputial opening, rendering the retraction of the foreskin difficult or impossible. By the end of the first year of life, the retraction of the foreskin is possible in 50% of boys, this percentage reaching 89% by the age of three. For children 6-7 years of age, the incidence of phimosis is 8%, reaching 1% for children aged 16-18 years. Amongst our cases phimosis was the most frequent anomaly identified, with a prevalence of 6.2%. This can be idiopathic or secondary to a retractile scar tissue (this is the case for brutal degloving, when the physiological adherences between the glans and foreskin are torn apart).

Phimosis is diagnosed by means of clinical examination. It exposes the child to the risk of recurrent urinary infections, especially when other factors are also involved (e. g. vesico-ureteral reflux). A very tight phimosis can also lead to micturition abnormalities with obstructive symptoms that may evolve to complete urinary retention.

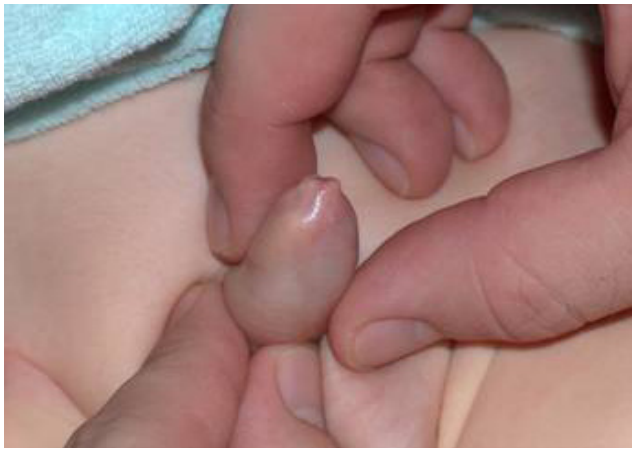

Figure 4. Congenital phimosis.

Hypospadias was found in 1.6% of the boys that we examined. The implications of this pathology are mainly esthetic and may lead to disorders regarding body image. Functional abnormalities are related to micturition, which may be altered by the presence of a possible associated urethral stenosis, and also to reproduction, the presence of hypospadias being able to interfere with fertilization [10].

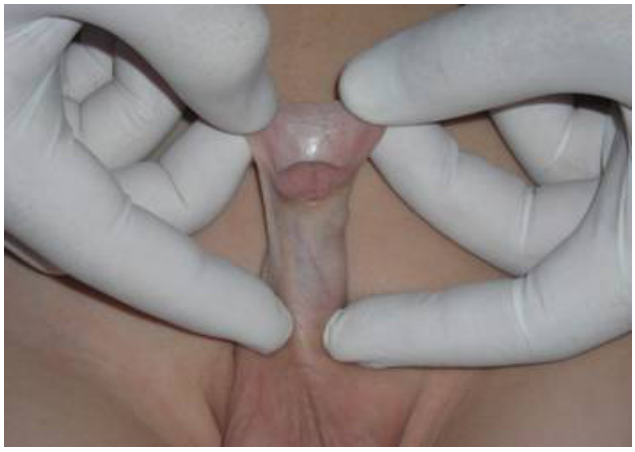

Figure 5. Coronal hypospadias.

We have established surgical indication for 43 patients (4.3%), namely the cases with cryptorchism, phimosis and hypospadias (figures 6, 7).

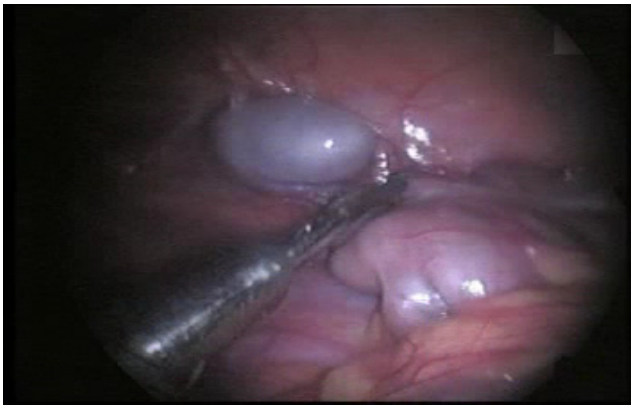

Figure 6. Laparoscopic surgery for cryptorchism with non-palpable testicle.

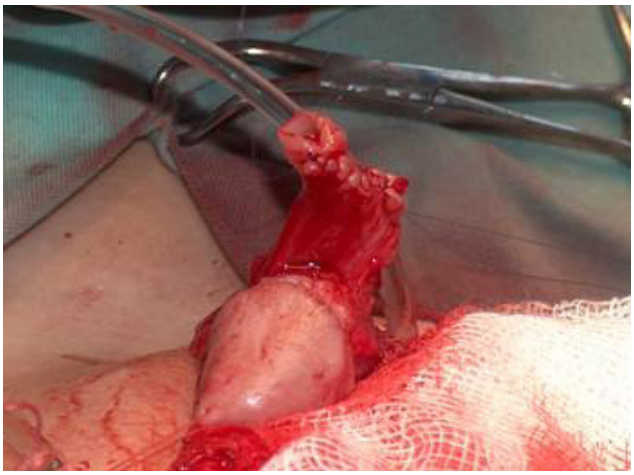

Figure 7. Preputial island flap urethroplasty for hypospadias.

Unlike clinical examination, ultrasound examination can identify urinary tract anomalies which may turn out to be much more severe and which may evolve more often towards renal failure: obstruction of the pyelo-ureteral junction, obstructive megaureter, and vesico-ureteral reflux.

Ultrasound examination diagnosed a third of the total abnormalities discovered, the rest being found upon clinical examination. Moreover, the ultrasound-diagnosed pathology did not require surgical treatment. The reason for this might be that the urinary tract anomalies were already discovered by the antenatal ultrasound examinations or by the pediatricians due to their severity or symptoms. In contrast, the genital anomalies may not be present at birth (the testicles may finish their descent at 2-3 months after birth, especially in preterm boys), so that they are missed by the medical examination after the boy is born.

CONCLUSIONS

The screening program for congenital uro-genital anomalies is effective and necessary. It required clinical and ultrasonographic evaluation of the urinary tract and external genital organs. This easily can be done by the family doctor or the pediatrician during routine medical examination. The high proportion of apparently healthy children that were having undiagnosed anomalies should prompt the family doctors or the pediatricians to examine the children in this respect during routine medical examination.

We believe that parents should also be instructed regarding the evaluation of the external genitalia of their children in order to notice possible anomalies that may require early surgical correction (e.g. checking the presence of testicles in the scrotum to prevent infertility and possible malignization in cryptorchism cases).

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