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Ultrasound screening of congenital urinary defects and its prospects in Slovakia

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The main purpose of the neonatal sonographic screening of the urinary system is to find defects that need to be resolved in the neonatal period or early infancy.

Objective: to establish the incidence and structure of congenital malformations in Slovakia and retrospectively determine the proportion of prenatal diagnostics in the revealing congenital malformations of the urinary system in the Slovak Republic during the last decade.

Materials and methods. In total 38,496 newborns were enrolled in the research, representing 66.5% of the total population of 57,969 newly-born children in 2017. In cooperation with the National Center for Health Information, we have found other statistical data.

Results. Grade 1 of the pelvicalyceal system dilatation according to Hofmann's classification occurred in 7%, grade 2 in 0.53%, grade 3 in 0.21%, and grade 4 in 0.1% of cases. One-sided agenesis occurred in about 0.1% of patients. Renal cysts of various etiologies revealed in about 0.05% of children. During the selected years, the proportion of the prenatal diagnosis of congenital developmental defects of the urinary system increased from 13.3% in 1995 to 29.1% in recent years (an average of 24.4%).

Conclusions. The incidence rate of congenital urinary malformations requiring nephrological and urological care and often intervention of urologist as early as in the neonatal period composes 31 out of 10,000 children. The prenatal diagnosis currently used in Slovakia finds only one quarter of congenital developmental defects of the urinary system. The high incidence rate, low-level coverage of prenatal diagnostic screening, history and physical examination, absence of a urinary infection in some forms and poor prognosis of the urinary pathology are clear arguments for the implementation of screening in all neonatal departments of the Slovak Republic.

Key words: incidence, newborn, kidneys, obstructive uropathy, screening, Slovakia, sonography.

Ультразвуковий скринінг вроджених дефектів сечової системи та його перспективи у Словаччині

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Основною метою неонатального ультразвукового скринінгу сечової системи є виявлення патології, яку необхідно усунути в неонатальному або ранньому дитячому віці.

Мета: встановити частоту та структуру вроджених вад розвитку сечової системи в Словаччині, а також ретроспективно визначити частку пренатальної діагностики.

Матеріали і методи. У дослідженні взяли участь 38 496 новонароджених, що становить 66,5% від загальної чисельності новонароджених (57 969 дітей) у 2017 році. У співпраці з національним центром медичної інформації були отримані й інші статистичні дані.

Результати. Дилатація чашково-мискової системи 1-го ступеня за класифікацією Гофманна становить 7%, 2 ступеня — 0,53%, 3 ступеня — 0,21% і 4 ступеня — 0,1%. Одностороння агенезія зустрічається приблизно у 0,1%. Ниркові кісти різної етіології зустрічаються приблизно у 0,05% дітей. За досліджувані роки частка пренатальної діагностики вроджених вад розвитку сечової системи збільшилася з 13,3% у 1995 році до 29,1% (у середньому 24,4%).

Висновки. Частота вроджених вад розвитку сечової системи, що вимагають нефрологічної та урологічної допомоги вже в неонатальному віці, становить 31:10 000. Наразі за допомогою пренатальної діагностики у Словаччині діагностується тільки чверть цих вад. Високий рівень захворюваності, низька частота виявлення на підставі пренатальної діагностики, анамнезу та фізикального огляду, відсутність у багатьох випадках інфекції сечової системи і поганий прогноз патології доводять необхідність скринінгу у всіх відділеннях новонароджених Словацької Республіки.

Ключові слова: захворюваність, нирки, новонароджений, обструктивна уропатія, скринінг, Словаччина, сонографія.

Ультразвуковой скрининг врожденных дефектов мочевой системы и его перспективы в Словакии

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Основной целью неонатального ультразвукового скрининга мочевой системы является обнаружение патологии, которую необходимо устранить в неонатальном или раннем младенческом возрасте.

Цель: установить частоту и структуру врожденных пороков развития мочевой системы в Словакии и также ретроспективно определить вклад пренатальной диагностики.

Материалы и методы. В исследовании приняли участие 38 496 новорожденных, что составляет 66,5% от общей численности новорожденных (57 969 детей) в 2017 году. В сотрудничестве с Национальным центром медицинской информации были получены и другие статистические данные.

Результаты. Дилатация чашечно-лоханочной системы 1-й степени согласно классификации Гофманна составляет 7%, 2 степени — 0,53%, 3 степени — 0,21% и 4 степени — 0,1%. Односторонняя агенезия встречается примерно у 0,1%. Почечные кисты различной этиологии встречаются примерно у 0,05% детей. За исследуемые годы доля пренатальной диагностики врожденных пороков развития мочевой системы увеличилась с 13,3% в 1995 году до 29,1% (в среднем 24,4%).

Выводы. Частота врожденных пороков развития мочевой системы, требующих нефрологической и урологической помощи уже в неонатальном возрасте, составляет 31:10 000. В настоящее время при помощи пренатальной диагностики в Словакии диагностируется только четверть этих пороков.

Высокий уровень заболеваемости, низкая выявляемость на основании пренатальной диагностики, анамнеза и физикального осмотра, отсутствие во многих случаях инфекции мочевой системы и плохой прогноз патологии доказывают необходимость скрининга во всех отделениях новорожденных Словацкой Республики.

Ключевые слова: заболеваемость, новорожденный, обструктивная уропатия, почки, скрининг, Словакия, сонография.

Introduction

The nationwide sonographic screening of congenital developmental anomalies of the kidneys in newborns is very important and carried out for a long time in the Slovak Republic. The beginnings of screening for congenital developmental anomalies of the kidneys date back to 1986 [8]. Although the screening is nationwide, there are some problems at various stages of its implementation, and the administrative problem is the long-term lack of legislation. Changes in diagnostic and therapeutic options and prenatal diagnostics give rise to new issues related to the screening strategy.

The first screening algorithms of congenital malformations of the kidneys were proposed in 1996 and in 2013; their author, Ol'ga Cervenova, M.D., associate professor, Ph.D in Medicine, was awarded the Crystal Wing for the promotion of the nationwide neonatal screening program [7,8]. The algorithms have been modified and adapted to the current situation with the growing amount of data from practice and statistics [2,4,5].

The main purpose of the neonatal sonographic screening of the urinary system is to find defects that need to be addressed in the neonatal period or early infancy (renal position anomalies, agenesis, dysplasia, obstructive uropathy dilation of the pelvicalyceal system according to Hofmann's classification (DHS), and cystic changes).

The current situation in the diagnosis of congenital malformations in Slovakia has not been analyzed for a relatively long time — 12 years [3,6]; therefore, we carried out a new analysis (mapping) throughout the territory of Slovakia to underline the importance of the screening of congenital kidney malformations, find problems related to the screening and data updating.

The proportion of prenatal diagnostics in the revealing congenital anomalies of the urinary system in the Slovak Republic has not yet been analyzed in more detail [1]. We used data from the National Health Information Center to find statistics on the proportion of prenatal diagnostics,

which were retrieved and provided from the National Register of Congenital Defects.

Materials and methods

Using a questionnaire survey, we collected statistical data from the neonatal centers; than the data were statistically processed; based on the collected data, we compared the incidence and structure of congenital urinary developmental defects with the statistical data from previous years.

The statistical analysis and processing of the collected data was carried out using SPSS for Windows version 21.0 [9].

The collected data was analyzed at the level of statistical description using the following statistical values: quantitative estimate (N), arithmetic mean (AM), standard deviation (SD), standard error of estimate (SE), median (Mdn), modus, sample variance, coefficients of skewness and kurtosis, maximum values.

The descriptive statistical indicators for the subject variables are summarized in Tables 1–3.

In total 38,496 newborns were involved in the research, accounting for 66.5% of the total population of 57,969 newly-born children in 2017 (Fig. 1). Assuming the most strictly established criteria (99% confidence level and 1% confidence interval), the minimum size of the study group

Table 1

Descriptive statistical indicators for continuous variables (2017)

Statistical Indicators	Number of newborns	Normal ultrasound
Number of centers	32	22
Mean	1203.00	1029.50
Standard error	134.897	124.344
Median	1097.00	971.50
Modus	296	269
Standard deviation	763.092	583.225
Skewness	1.535	1.026
Kurtosis	3.345	2.117
Range	3504	2458
Minimum	296	269
Maximum	3800	2727
Sum	38496	22649

Descriptive statistical indicators for continuous variables (2017)

Statistical Indicators	DDS Grade I	DDS Grade II	DDS Grade III	DDS Grade IV
Number of centers	24	24	24	27
Mean	76.63	5.79	2.38	1.15
Standard error	20.053	1.144	0.446	0.281
Median	38.00	3.00	2.00	1.00
Modus	25	2	1	0
Standard deviation	98.238	5.603	2.183	1.460
Skewness	2.764	1.078	1.141	1.326
Kurtosis	9.202	0.114	0.841	0.866
Range	453	19	8	5
Minimum	1	0	0	0
Maximum	454	19	8	5

Table 2

Descriptive statistical indicators for continuous variables (2017)

Statistical Indicators	Agenesis	Suprarenal hemorrhage	Other (Cysts and other)
Number of centers	27	25	18
Mean	1.33	2.76	4.67
Standard error	0.256	0.466	1.221
Median	1.00	2.00	2.50
Modus	1	0	1
Standard deviation	1.330	2.332	5.179
Skewness	1.871	0.722	1.564
Kurtosis	5.114	0.327	2.138
Range	6	9	19
Minimum	0	0	0
Maximum	6	9	19

Table 3

should be 57,969 persons, the results of which can be generalized to a total population of 12,930 individuals. Since our study group included 38,496 individuals, we can generalize (extrapolate) the results for the entire population.

Furthermore, in cooperation with the National Center for Health Information (NCHI), we have obtained statistical data on the proportion of prenatal diagnostics in revealing the congenital developmental defects of the urinary system for the

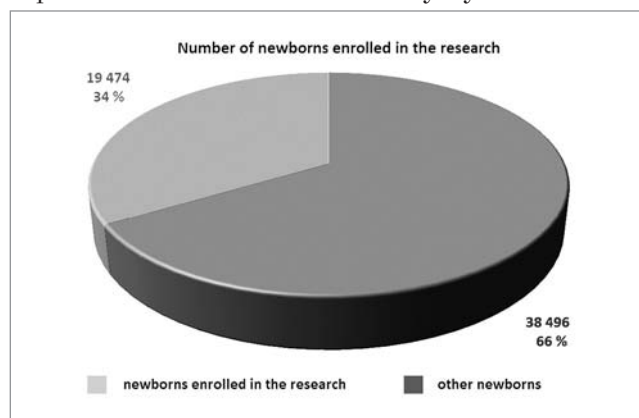


Fig. 1. Number of newborns enrolled in the research

years 1995, 2000, 2005, 2008 and 2013–2016. Data for the selected years was retrieved and provided from the National Register of Congenital Defects [10]. The statistics for 2017 and 2018 are not yet available, as data processing begins after the end of the calendar year and according to the World Health Organization (WHO), for children who were born in the current year, the diagnosis of the congenital developmental defects and reporting of the congenital defects is postponed upon reaching the child 12 months.

Results

In our study, we found that the number of neonatal departments decreased from 65 (in 1996) to 54 and we received statistical data from 33 (61.11%) neonatal departments.

According to our findings, ultrasound renal screening was performed in 100% neonatal departments that participated in our study.

The equipment of all study sites is suitable; the departments are provided with the modern ultrasound apparatuses.

However, 15 (27%) of neonatal centers did not record accurate statistics; some centers reported only grade 3 and 4 of the pelvicalyceal system dilation according to Hofmann's classification.

In 18 (33%) neonatal centers, the screening was performed by a radiologist or pediatrician because they had insufficient number of physicians or trained staff, and one department reported about the insurance company work requirement.

Some doctors emphasized that screening was not a mandatory examination and, therefore, it was done out of tradition, however they were trying to do it honestly.

Grade 1 of the pelvicalyceal system dilation was found in 4,057 (7%) newborns, grade 2 in 307 (0.53%), grade 3 in 121 (0.21%), and grade

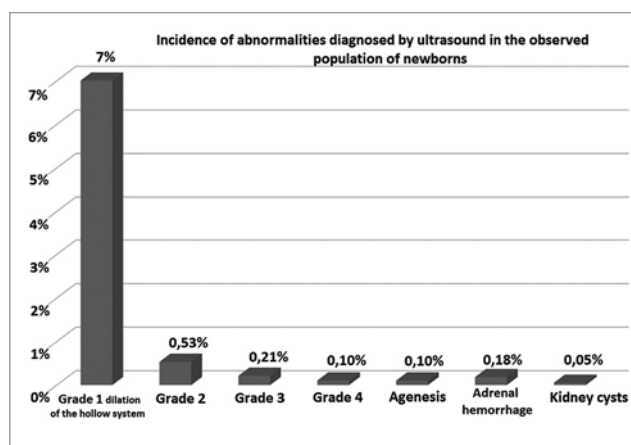


Fig. 2. Incidence of abnormalities diagnosed by ultrasound in the observed population of newborns

Table 4

Descriptive statistical indicators for continuous variables

Incidence of abnormalities	2017	2006
Grade I DHS	7%	6%
Grade II DHS	0.53%	1%
Grade III DHS	0.21%	0.30%
Grade IV DHS	0.10%	0.10%
Agenesis	0.10%	0.14%
Suprarenal hemorrhage	0.18%	
Other (such as cysts)	0.05%	0.05%

Table 5

Percentage proportion of prenatally diagnosed congenital urinary malformations

Year	Proportion (%) of prenatally diagnosed congenital defects of the urinary system (ICD Diagnosis: Q60-Q64)	mean, %
2013	23.0	24.4
2014	21.3	
2015	24.3	
2016	29.1	

Source: National Register of Congenital Defects, NCHI.

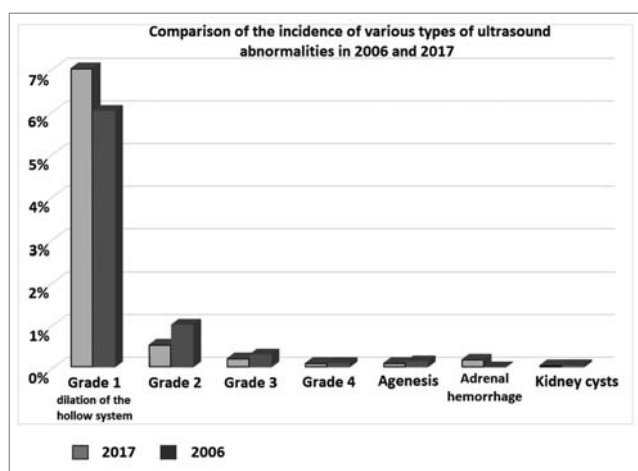
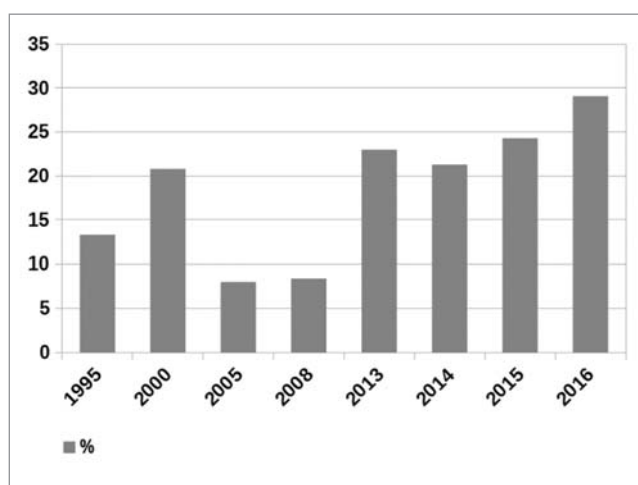


Fig. 3. Comparison of the incidence of various types of abnormalities revealed ultrasonically in 2006 and 2017



Source: National Register of Congenital Defects, NCHI.

Fig. 4. Proportion (%) of prenatal diagnosis of congenital urinary malformations in the period of 1995–2016

4 in 58 (0.1%) children. Unilateral agenesis occurred in about 0.1% (58 children). Kidney cysts of various etiologies occurred in 29 (0.05%) children (Figure 2, Table 4).

According to our findings, the structure of pelvicalyceal system dilations is not significantly different in 2017 versus the period of 1996–2006 (Fig. 3) [3,10].

The head physicians from 19 (almost 58%) neonatal departments reported that they have identical statistical data each year.

The study of prenatal diagnosis was completed and its results are shown in graphs and tables for better presentation. All congenital malformations of the urinary system, including combined defects, comprised in the statistics. The percentage of the prenatal diagnosis of the congenital urinary malformations is shown in Fig. 4.

The percentage of prenatally diagnosed congenital malformations of the urinary system in the period of 2013–2016 is provided in Table 5.

As we can see, the percentage of prenatally diagnosed congenital defects of the urinary system in recent years is only 24.4.

Since 1995, there has been an almost doubling of the prenatal diagnosis of congenital defects of the urinary system, which is a significant development.

According to the NCHI staff, we found out that the problem of registering congenital malformation is that with combined congenital defects, there is not 100% certainty, which congenital defects are prenatally diagnosed. This depends on the severity and the ability to visualize multiple congenital defects. Prenatal diagnosis of several congenital malformations indicates a higher level of prenatal diagnosis of combined congenital malformations, but it is not clear to what degree the congenital defects of the urinary system are visualized.

In any case, the number of prenatally diagnosed congenital malformations of the urinary system is increasing with respect only to the defects of the urinary system. The reporting modification to clarify these data in the near future is not yet possible since the reporting system is imperfect and the statistical data processing for 2018 is not yet complete.

Moreover, until 2012, the statistics on congenital urinary defects also included those determined in fetuses after maternal induced abortions, distorting the statistical data. Since 2012, statistics on abortions are recorded not in congenital defects, but in the abortion statistics reports.

Discussion and conclusion

The neonatal ultrasound screening of congenital malformations of the kidneys is the first stage in the diagnosis of severe kidney disease, the treatment of which then requires multidisciplinary collaboration, especially first-contact doctors, nephrologists and urologists.

According to our statistical data, in the Slovak Republic, the congenital urinary defects are diagnosed only in **one fourth** of children in the prenatal period. The level of prenatal screening in the different regions of Slovakia varies, as well as among the doctors themselves. Currently, the real and greatest advantage of a prenatal obstructive uropathy diagnosis is the possibility for early post-natal follow-up and appropriate treatment. The improvement of prenatal diagnosis in recent years has been an important step.

Due to the post-natal screening, we currently determine these anomalies before the clinical manifestations. According to our findings, the structure of the pelvicalyceal system dilations does not differ significantly compared to the data of 1996–2006. Severe congenital malformations of the urinary tract, requiring nephrological and uro-

logical care, and often intervention of urologist in infancy (particularly, the severe dilations of grade 3 and 4 according to Hoffmann's classification), affect about **31 out of 10,000** children.

The high incidence rate, low-level coverage of prenatal diagnostic screening, history and physical examination, absence of a urinary infection in some forms and poor prognosis of the urinary pathology are **clear arguments for the implementation of screening in all neonatal departments of the Slovak Republic** and congenital kidney malformations are still a pressing problem.

There are persisting problems with the training of personnel; there is a lack of doctors in the pediatric and neonatal care sector who would be capable of regularly carrying out this screening. Also, to avoid any misunderstandings, the ultrasound screening of congenital kidney developmental defects should be included in the group of recommended or mandatory screening examinations in the form of the expert guidance by the Ministry of Health or the guideline by the corresponding society of specialists to close the gap with the missing legislation.

The author does not declare any conflicts of interest.

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