Congenital malformations of the urogenital system in the Republic Sakha (Yakutia)

I.R. Egorova, A.N. Nogovitsyna, N.R. Maksimova, A.L. Sukhomyasova

Yakutsk Science Center of Complex Medical Problems of Russian Academy of Medical Sciences, Yakutsk, the Republic Sakha (Yakutia), the Russian Federation

The present study analyzes the data of the National monitoring of congenital malformations for 2000-2009 and for the first time the frequency, structure of malformations of the urogenital system of newborns and aborted fetuses in Yakutia are determined. The estimation of the level of effectiveness of the prevention of congenital malformations is given.

Keywords: monitoring, congenital malformations, urogenital system, fetus, newborn.

Introduction. Territorial registers and monitoring of congenital anomalies (CA) are of great significance for estimating showings of genetic cargo of the population. The congenital anomalies are considered to be one of the causes of prenatal mortality rate [3]. According to some studies conducted in different countries, 25-30% of all prenatal losses are caused by anatomic defects. The maximum rate of congenital defects (to 80-85%) is observed at early stages of prenatal development that has been revealed while studying materials of spontaneous abortions [8]. Among deadborns CA occur in 15-20% of cases. Within 1st year of life 25% of all cases of mortality are caused by congenital anomalies [4].

Congenital anomalies of urogenital system (UGS) appear to be serious medical-social problem as they cause the development of urinary tract infection (40-80%), up to the development of chronic renal failure (88%) [10, 11]. The congenital renal anomalies among children are not only common, but also have growing tendency, rating 30-50% of all congenital anomalies [2, 6, 13]. CA of UGS refer to the list of the most frequent congenital anomalies which are included in the International defects register which are subject to genetic monitoring (International Birth Defects Monitoring System, Eurocat). According to the incorporated register EUROCAT and the register of the Russian Federation for 2000-2010 the defects of UGS occupy 3rd place (15,17 and 17,18% accordingly) in the structure of leading congenital anomalies. In the structure of UGS pathology hypospadias is noted as the most prevalent (41, 98%), it followed by kryptorhism (21,56%), and then congenital defects of pyelectasia passage (13,45%) [3].

The genesis of congenital anomalies is mainly connected with chromosomal aberrations,

however, they can be manifested by rare monogenic illnesses (approximately in 2% of cases). More than 80% of CA have polygene nature and are connected with teratogen affection of the environment [1]. Most researchers consider that such exterior factors have adverse effect to the formation of primary kidney, leading to chronic hypoxia of nephritic fabric and development of various fetus system failures that can be manifested in various renal pathology [2, 9, 12, 14]. General principles of formation of urinary system defects are connected with histogenesis failure at cellular reproduction level, cellular growth, migration and differentiation, formations of intercellular and intrafabric cooperations. The failure of any links in development of urinary system can lead to defect formation [15].

Thus, because of high incidence rate of the urogenital system and its great influence to the structure of infantile death rate, infantile morbidity and physical disability it is necessary to study the given problem more thoroughly.

Materials and methods: 140,018 data about newborns, a database of the Republican register of congenital anomalies, genetic cards of pregnant women, the form №025-11/at-98 on an infant with congenital anomalies. The analysis of data has been conducted by means of program Statistika 6,0.

Results and discussion: The analysis of Republican monitoring data on anomalies shows that for 10 years 289 cases of urogenital system anomalies (UGS) at newborns and fetus, including 192 isolated anomalies and 97 cases as a part of plural anomalies have been registered. Besides, 236 cases of pyelectasia have been noted: unilateral - 185 (Yakutsk - 115, in rural areas - 70), bilateral - 51 (Yakutsk - 26, in rural areas - 25). Mid-annual frequency of urogenital system defect at newborns and eliminated fetuses has made 0,14%. The highest incidence rate of UGS was marked in 2006-2007, thus the frequency of isolated defects has amounted 0,25%, and in composition MCD (multiple congenital defects) - 0,1% (fig. 1).

Within the studied period 164 newborns with UGS isolated anomalies were revealed, and 28 fetuses were eliminated subject to medical outcomes (Tab. 1). Fetuses with CA UGS were eliminated with parental consent, and basically, due to bilateral process (bilateral renal agenesia, bilateral renal hypoplasia). The efficiency of preventive measures was estimated in 14,5%. In the structure of UGS isolated defects at newborns and interrupted fetuses hydronephrotic renal transformation (Q 62.0) - 0,07% and infantile polycystic kidney (Q 61.1) - 0,02% (Tab. 1). On the third place unilateral renal agenesia (Q 60.0) - 0,01% was noted at newborns, and at fetus there were other specified UGS anomalies. The general frequency of UGS isolated anomalies at newborns has amounted 0,12%, and at interrupted fetuses - 0,02%.

UGS anomalies in composition of congenital anomalies have been verified in 97 cases: 72 newborns (74,2%), 25 eliminated fetuses (25,7%) (Tab. 2). In the general structure of UGS anomalies in composition of multiple anomalies among newborns (0,01%) the leading positions are taken by hydronephrosis (0,02%) and unilateral renal agenesia (0,02%), the second place - infantile polycystic kidney (0,01%). There was timely detection and elimination of fetuses with infantile polycystic kidney - 7, with unilateral renal agenesia - 5, with bilateral renal hypoplasia – 4. Newborns with UGS severe and rebellious anomalies in composition of MCD (bilateral renal agenesia - 3, infantile polycystic kidney - 4, bladder extrophy - 2), as well as with hydronephrosis and unilateral renal agenesia ($p<0,05$) were significantly born in rural areas than in Yakutsk. The frequency of UGS anomalies in composition of multiple congenital anomalies at newborns has been 0,05%, and 0,02% at interrupted fetuses accordingly. The general frequency of UGS congenital anomalies in composition of MCD has been 0,07%, and after preventive measures it has decreased to 0,05% at newborns.

In connection with lack of some data the study of women's anamnesis with indications on live or dead birth as well as a fetus with CA UGS on genetic cards has not revealed objective risk factors of congenital anomalies formation.

The conclusion. Thus, unilateral renal agenesia and hydronephrosis appear to be the most frequent renal anomalies at newborns, the latter can undergo to surgical correction depending on injury level. Authentically high frequency of birth rate cases with CA UGS including rebellious anomalies in rural areas is caused by insufficient preventive actions.

Children with pre-natal revealed pathology of urinary system should undergo dynamic supervision at early age for early diagnostics of UGS diseases, preventive measures of urinary tract infection, nephrosclerotic formation and nephritic insufficiency [5]. To conduct suitable prophylaxis of congenital anomalies it is necessary to improve the level, possibilities and quality of diagnostics, and also to use «Instructions on CA phenotypical manifestations» [7] and multimedia directory system «Congenital anomalies» [4].

Figure 1

Frequency of isolated defects of urogenital system and in composition of multiple congenital defects at newborns and fetuses for 2000-2009

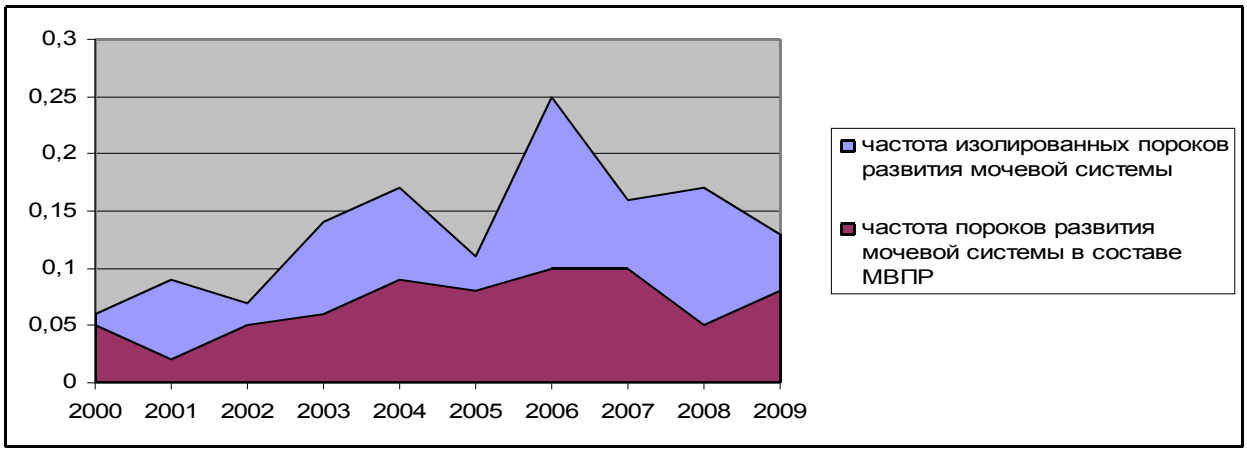

Table 1

Isolated congenital anomalies of urogenital system in Republic Sakha (Yakutia) for 2000-2009

Defect type	Live birth (newborns)				Eliminated (fetuses)				Total n	Frequency (%)
	Yakutsk n	Districts n	Total n	Frequency (%)	Yakutsk n	Districts n	Total n	Frequency (%)		
Unilateral renal agenesia	13	4	17	0,01	-	-	-	-	17	0,01
Bilateral renal agenesia	-	-	-	-	2	-	2	0,001	2	0,001
Unilateral renal hypoplasia	2	4	6	0,004	-	-	-	-	6	0,004
Bilateral renal hypoplasia	-	-	-	-	1	-	1	0,001	1	0,001
Single renal cyst	1	1	2	0,001	-	-	-	-	2	0,001
Infantile polycystic kidney	7	13	20	0,01	3	3	6	0,004	26	0,02
Unclassified polycystic kidney	-	1	1	0,001	-	-	-	-	1	0,001
Renal pelvis passage failure and ureter anomaly	-	-	-	-	1	-	1	0,001	1	0,001
Hydronephrosis	44	44	88	0,06	7	4	11	0,008	99	0,07
Ureter atresia and stenosis	1	-	1	0,001	-	-	-	-	1	0,001
Megaloureter	-	1	1	0,001	-	-	-	-	1	0,001
Reflux bladder ureter	3	1	4	0,003	-	-	-	-	4	0,003
Accessory kidney	2	-	2	0,001	-	-	-	-	2	0,001
Merged, lobular and U-shape kidney	3	5	8	0,006	-	1	1	0,001	9	0,006
Ectopic kidney	3	5	8	0,006	-	-	-	-	8	0,006
Unclassified renal anomaly	1	-	1	0,001	-	-	-	-	1	0,001
Posterior urethral valves	1	1	2	0,001	-	-	-	-	2	0,001
Other bladder and urethra anomalies	-	-	-	-	1	-	1	0,001	1	0,001
Other classified urinary system anomalies	1	-	1	0,001	2	1	3	0,002	4	0,003
Unclassified urinary system anomaly	-	2	2	0,001	-	-	-	-	2	0,001
Total:	82	82	164	0,12	19	9	28	0,02	192	0,14

Table 2

Urinary system anomalies in composition of multiple congenital anomalies in Republic Sakha (Yakutia) for 2000-2009

Diagnosis	Live birth (newborns)				Eliminated (fetuses)				Total n	Frequency (%)
	Yakutsk n	Districts n	Total n	Frequency (%)	Yakutsk n	Districts n	Total n	Frequency (%)		
Agenesia and other reduction renal defects	-	-	-	-	1	-	1	-	1	0,001
Unilateral renal agenesia	6	11	17	0,01	2	3	5	0,004	22	0,02
Bilateral renal agenesia	-	3	3	0,002	2	1	3	0,002	6	0,004
Unilateral renal hypoplasia	7	1	8	0,006	-	-	-	-	8	0,006
Bilateral renal hypoplasia	1	-	1	0,001	3	1	4	0,003	5	0,004
Infantile polycystic kidney	-	4	4	0,003	3	4	7	0,005	11	0,01
Unclassified polycystic kidney	-	1	1	0,001	-	-	-	-	1	0,001
Renal dysplasia	-	1	1	0,001	-	-	-	-	1	0,001
Hydronephrosis	7	12	19	0,01	1	1	2	0,002	21	0,02
Ureter atresia and stenosis	-	1	1	0,001	1	-	1	0,001	2	0,001
Ureter agenesia	-	1	1	0,001	-	-	-	-	1	0,001
Accessory kidney	1	2	3	0,002	-	-	-	-	3	0,002
Merged, lobular and U shape kidney	3	4	7	0,005	2	-	2	0,001	9	0,01
Ectopic kidney	2	1	3	0,002	-	-	-	-	3	0,002
Bladder extrophy	-	2	2	0,001	-	-	-	-	2	0,001
Other types of ureter and bladder neck atresia and stenosis	-	1	1	0,001	-	-	-	-	1	0,001
Total:	27	45	72	0,05	15	10	25	0,02	97	0,07

References

Veltishchev J.E. Infantile inability: medical and social aspects, preventive measures. The lecture for physicians / J.E. Veltishchev, D.I. Zelinsky // Russ. reporter of prenatal. and pediatrist. The appendix 2000. – 68 p.

Geld V.G. Diagnostics of urogenital system defects at newborns and infants / V.G. Geld, G.I. Kuzovleva // Pediatrics. - 2006. – № 1. – P. 87-94.

Demikova N.S. Congenital anomalies in regions of the Russian Federation (monitoring showings for 2000-2010) / N.S. Demikova, A.S. Lapina / Russ. reporter of prenatal. and podiatrist. – 2012. – № 2. – P. 91-98.

Demikova N.S. Directory system on congenital anomalies in medical practice and formation / N.S. Demikova, A.S. Lapina, A.N. Putintsev, N.N. Shmeleva / VIT. – 2007. – № 6. – p. 33-36.

Egorov I.R. Structure of microbe-inflammatory diseases of urinary system at infants with prenatal revealed changes in kidneys / I.R. Egorov, E.I. Kondratyev, A.A. Terentyev G.N. Jankina // The Kuban scientific medical bulletin. – 2010. – № 7. – P. 178-182.

Ignatov M.S. Prevalence of urinary system diseases at children / M.S. Ignatov // Russ. reporter of prenatal. and pediatrist. – 2000. – № 1. - P. 24-29.

The Instruction on phenotype description at infants with congenital anomalies / L.Z. Kazantseva, A.I. Klembovskiy, N.S. Demikova etc. MH RF 2001; 22 p.

Muller R.F. Emery's Elements of Medical Genetics / R.F. Muller, I.D. Jong. – Edinburgh: Churchill Livingston. – 2001. – 256 p.

Makovetskaja G.A. To the question on kidney diseases at newborns and first month infants / G.A. Makovetskaja, J.I. Mazuk, T.V. Kozlova // Nephrology and dialysis. – 2000. – T. 2. – № 1. - P. 51-54.

Papayan A.V. Neonatal nephrology. The management / A.V. Papayan, I.S. Styazhkina. – StPb: Peter, 2002. – 434 p.

Papayan A.V. Features of disease state and treatment of urinary system infections at infants / A.V. Papayan, I.S. Styazhkina // Russ. reporter of prenatal. and pediatrist. – 2002. – № 4. – P. 47-51.

Pasechnik I.N. Mechanisms of activated oxygen damaging action on biological structures of patients in critical conditions / I.N. Pasechnik // Reporter of intensive therapy. – 2001. – № 4. – P. 4-9.

Tsaregorodtseva A.D. Urinary system diseases at children / A.D. Tsaregorodtseva, M.S. Ignatov // Russ. reporter of prenatal. and pediatrics. – 2001. – № 4. – P. 25-28.

Chugunova O.L. Urinary system diseases at newborns / O.L. Chugunova // Russ. reporter of prenatal. and pediatrist. – 2006. – № 2. – P. 18-23.

Shabalov N.P. Neonatology / N.P. Shabalov – StPb: Peter, T. 1, 1997. – 555 p.

Contacts

Egorova Iraida Rufovna – PHD, MD, senior researcher Yakut Scientific center of Siberian Department of Russian academy of Medical Sciences, Yakutsk, the Republic Sakha (Yakutia), the Russian Federation

Nogovitsyna Anna Nikolaevna – PHD, MD, geneticist of the Republican hospital №1- National Medical Center, head of laboratory of Siberian Department of Russian academy of Medical Sciences, Yakutsk, the Republic Sakha (Yakutia), the Russian Federation, e-mail: **nogovan@yandex.ru**

Maksimova Nadezhda Romanovna – PHD, MD, geneticist of the Republican hospital №1- National Medical Center, head of laboratory Siberian Department of Russian academy of Medical Sciences, Yakutsk, the Republic Sakha (Yakutia), the Russian Federation.

Sukhomyasova Aytalina Lukinychna – PHD, MD, geneticist of the Republican hospital №1- National Medical Center, head of laboratory Siberian Department of Russian academy of Medical Sciences, Yakutsk, the Republic Sakha (Yakutia), the Russian Federation, e-mail: **aitalinaS@yandex.ru**