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CHARACTERISTICS OF CONGENITAL PATHOLOGY WITH INHERITED AND MULTIFACTORIAL NATURE IN CHILDREN OF KYIV REGION

Abstract. *In the process of conducting epidemiological studies congenital pathology with inherited and multifactorial nature of children in Kiev region, 2010-2015, was characterized. The cases of diagnosis of newborns and individuals whose pathology was found for the first time beyond the neonatal period are reviewed.*

Key words: *congenital developmental defects; hereditary pathology; primary prophylaxis.*

Introduction. Among newborns in Ukraine a certain fluctuation range of genetic diseases is found that can be associated with various causes, and the following ones may occupy not the last position among them: availability of medical-genetic aid, quality and fullness of diagnostics, accurate registration of pathology, effect of primary and secondary prevention measures [1].

Therefore, the **objective** of the study was assessment of congenital pathology among newborns and individuals whose pathology was first found beyond the neonatal period.

Materials and methods. The objects of the study were cases of congenital and inherited pathology in newborns, in patients with the diagnosis first made among the individuals older than one year. To characterize congenital pathology in newborns (507 cases) and beyond the neonatal period (953 cases), the data of hospital registers of Kyiv region from the establishment "Kyiv Regional Center of Health Care of Mother and Child" during 2010-2015 were used. Methods of investigation: epidemiological, statistical.

Results. First, congenital pathology among newborns have been characterized. Its structure during 2009-2015 is indicative of prevailing congenital developmental defects (CDD) of the circulatory system (Q 20-28), who constituted one third of the registered CDD (30,18±2,04 %). The second position belonged to CDD of the osseous-muscular system (Q 65-79) - (24,85±1,92 %), and

the third one – CDD of the gonads (Q 50-56), (17,75±1,70 %).

Every fifteenth defect was cleft lip or palate (Q 35-37) (6,51±1,10 %), practically every twelfth CDD referred to the category of "CDD of the digestive organs Q 38-45" (3,75±0,84 %) or "chromosome abnormalities not classified in other groups Q 90-99" (4,14±0,88 %), every thirtieth multifactorial CDD (MCDD) including defects of several classes (3,16±0,78 %), CDD of the urinary system, Q 60-64 (2,96±0,75 %) and CDD included into other CDD, Q 80-89 (2,96±0,75 %). Specific gravity of CDD of the nervous system (Q 00-07), CDD of the eye, ear, face (Q 10-18) and respiratory CDD (Q 30-34) was 1,78±0,59 %, 1,58±0,55 % and 0,39±0,28 % respectively.

To characterize congenital pathology among newborns several groups of analysis were considered to be reasonable to differentiate:

- I – newborns whose parents were born in the region of residing;
- II – newborns whose parents were born in different districts of Kyiv region;
- III – newborns whose parents were born above the borders of the region.

The structure of congenital pathology among newborns whose parents were born and resided within the borders of one district of Kyiv region is indicative of prevailing CDD of the circulatory system (Q 20-28), constituting one fifth and one seventh part of all the registered CDD (18,34±1,72 % - for mothers and 13,21±1,50 % - for fathers).

CDD and deformities of the osseous-muscular system (Q 65-79) were on the second position – every seventh defect ($14,20 \pm 1,55$ % and $13,61 \pm 1,52$ % respectively), CDD of the gonads – on the third position (Q 50-56), ($9,47 \pm 1,30$ % and $9,47 \pm 1,30$ % respectively).

Every twentieth and thirtieth defect was cleft lip or palate (Q 35-37) ($4,73 \pm 0,94$ % - for mothers and $2,76 \pm 0,73$ % - for fathers), practically every forty-second and every fiftieth CDD referred to the category «CDD of the digestive organs Q 38-45» ($2,37 \pm 0,68$ % and $1,97 \pm 0,62$ % respectively) or every fiftieth or forty-ninth CDD referred to «chromosome abnormalities not classified in other groups Q 90-99» ($1,78 \pm 0,59$ % and $2,56 \pm 0,70$ % respectively). Specific gravity of the rest CDD was not considerable (up to 2 %).

The structure of CDD among newborns included II group of the analysis is indicative of prevailing CDD of the circulatory system (Q 20-28), constituting the thirtieth and twentieth part of all the registered CDD ($2,76 \pm 0,73$ % - for mothers and $4,93 \pm 0,96$ % - for fathers). The second position was occupied by CDD of the gonads (Q 50-56) and CDD and deformities of the osseous-muscular system (Q 65-79) – where every fiftieth defect ($2,17 \pm 0,65$ % and $2,37 \pm 0,68$ %; $1,97 \pm 0,62$ % and $2,17 \pm 0,65$ % respectively), the third one – by MCDD, - practically every hundredth defect among newborns ($0,59 \pm 0,34$ % and $0,79 \pm 0,39$ % respectively). The rest of CDD were less than 1%, and minimal specific gravity was found among respiratory CDD (0,00 % both for mothers and fathers).

The structure of congenital pathology among newborns of III group is indicative of prevailing CDD of the circulatory system (Q 20-28), constituting the thirteenth part of all the registered CDD ($7,50 \pm 1,17$ % among mothers). CDD and deformities of the osseous-muscular system (Q 65-79) were on the second position – every fourteenth defect ($7,10 \pm 1,14$ %), on the third position – CDD of the gonads (Q 50-56), - practically every twentieth defect among newborns ($5,13 \pm 0,98$ %). The rest of CDD were less than 1%, and minimal specific gravity was found among CDD of the eye, ear, face and neck (0,00 %).

The structure of congenital pathology among newborns of III group of the study is indicative of

prevailing CDD of the circulatory system (Q 20-28), constituting the thirteenth part of all the registered CDD ($7,69 \pm 1,18$ % among fathers). The second position is occupied by CDD and deformities of the osseous-muscular system (Q 65-79) – every fourteenth defect ($6,71 \pm 1,11$ %), the third one – CDD of the gonads (Q 50-56), - practically every twenty-fifth defect among newborns ($3,94 \pm 0,86$ %). Minimal specific gravity was found among respiratory CDD (0,20 %), and specific gravity of the rest CDD was less than 1%.

While characterizing individuals whose pathology was found beyond the neonatal period it was found that the structure of congenital pathology among those children during 2010-2015 was prevailing CDD included into the group “other CDD” (Q 80-89), constituting one third of all the registered CDD ($30,85 \pm 1,50$ %). The second position is occupied by CDD and deformities of the osseous-muscular system (Q 65-79) – every fourth defect ($26,97 \pm 1,44$ %), the third position – chromosome abnormalities not classified in other groups (Q 90-99), practically every seventh defect among newborns ($14,48 \pm 1,14$ %).

Every eleventh defect was MCDD (including CDD of several classes) ($8,60 \pm 0,91$ %), practically every thirteenth CDD referred to the category «CDD of the nervous system Q 00-07» ($7,66 \pm 0,86$ %), every fortieth – CDD of the eye, ear, face, neck (Q 10-18) ($2,52 \pm 0,51$ %), every fiftieth – phenylketonuria (FKU, E 70.0) ($2,20 \pm 0,48$ %), CDD of the circulatory system, Q 20-28 ($2,10 \pm 0,46$ %) and CDD of the gonads, Q 50-56 ($1,89 \pm 0,44$ %). Specific gravity of hypothyroidism (E 03.1), CDD of the digestive organs (Q 38-45), CDD of the urinary system (Q 60-64) and cleft lip and palate (Q 35-37) were $1,15 \pm 0,35$ %, $0,73 \pm 0,28$ %, $0,52 \pm 0,23$ % and $0,31 \pm 0,18$ % respectively.

Concerning the characteristics of a middle age as to the making diagnosis of patients (Table), syndrome diagnosis were found to be made at the latest stages (other CDD, Q 80-89) ($14,69 \pm 0,58$ years). Hypothyroidism was on the second position, E 03.1 ($12,84 \pm 3,27$ years), on the third one – CDD of the eye, ear, face and neck Q 10-18 ($11,54 \pm 2,02$ years).

Manifestation of a number of CDD occurs beyond the neonatal period. As it is evidenced by previously conducted studies and proved by present findings, special attention should be paid

Table
An average age to make the diagnosis of patients according to the classes of diseases (ICD -10), Kyiv region, 2010-2015.

Classification according to ICD-10	Average age according to nosology, years	Minimal value of age of making diagnosis, years	Maximal value of age of making diagnosis, years
Q 00-07	7,13±0,87	1,00	31,00
Q 10-18	11,54±2,02	1,00	40,00
Q 20-28	10,79±1,11	1,42	17,00
Q 35-37	10,67±2,96	5,00	15,00
Q 38-45	9,86±0,94	7,00	13,00
Q 50-56	5,94±1,21	1,00	14,00
Q 60-64	6,05±1,58	2,25	11,00
Q 65-79	9,12±0,44	1,00	44,00
Q 80-89	14,69±0,58	1,00	44,00
Q 90-99	9,37±0,61	1,00	30,00
E 03.1 (hypothyroidism)	12,84±3,27	1,08	34,00
E 70.0 (FKU)	10,58±1,71	1,00	28,00
MCDD	9,47±0,95	1,00	48,00
All CDD	10,85±0,28	1,00	48,00

to FKU and hypothyroidism. Their timely detection will enable to indicate the essential course of treatment, since children with such diagnoses made after one year of their life, are subjected to irreversible disability.

Conclusions. Analysis of the hospital register of patients with the first made diagnosis of congenital pathology beyond the neonatal period during 2010-2015 was indicative of prevailing CDD

included into the group “other CDD” (Q 80-89), constituting one third of all the registered CDD (30,85±1,50 %). The second position belonged to CDD and deformities of the osseous-muscular system (Q 65-79) – every fourth defect (26,97±1,44 %), the third position – chromosome abnormalities not classified in other groups (Q 90-99) – (14,48±1,14%).

An average age of making the diagnosis of patients according to the classes of diseases is indicative of inefficacy of the screening system of FKU and hypothyroidism. Thus, the diagnosis of FKU was first made even at the age of 28 (a mean value – 10,58±1,71), hypothyroidism – at 34 (a means value – 12,84±3,27). Syndrome diagnoses were made at the latest period of life (at 14,69±0,58), the diagnoses of congenital defects of the eye, ear, face (11,54±2,02), congenital developmental defects of the circulatory system (10,79±1,11), and cleft lip or palate (10,67±2,96).

Prospects of further studies. The studies conducted enable to improve the system of prevention of congenital pathology at an individual level when a family doctor is performing his professional duties, and in case of specification of preventive measures; to reduce social and economic consequences.

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