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Congenital malformations in children up to 1 year of age in the autopsy material from the Department of Pathomorphology, Medical Academy in Lublin in the years 1984 – 1998

Wady wrodzone u dzieci do pierwszego roku życia w materiale autopsyjnym Katedry i Zakładu Patomorfologii Akademii Medycznej w Lublinie w latach 1984 – 1998

The morphologic anomalies that occur during intrauterine development from fertilization to the end of organogenesis are called congenital malformations (16). It is supposed that the incidence of congenital malformations among neonates and infants is about 4% of all live births and stillbirths (12). They are one of the major causes of death in this period of life (1, 7). Etiology of congenital malformations is complex and still poorly understood (12, 13). In 30% of cases they are thought to be genetic in origin (e.g. chromosomal aberrations, gene mutations). Environmental factors (as for instance irradiation, chemicals, drugs, hormones, malnutrition, hypoxy, maternal infections) induce malformations in about 10% of cases. Other 60% of cases are multifactorial in origin (genetic and environmental) (13, 16). In the opinion of some investigators the cause of the most congenital malformations is the hereditary genetic predestination, and the environmental factors only help them to express (16).

In the light of data on the progressive environmental degradation connected with industralization, urbanization, development of motor transport, excessive utilization of chemicals in agriculture and after the damage of atomic power plant in Tschamobyl in 1986, that is after the general increase of environmental teratogens (17, 18, 19), we decided to analyse congenital malformations in children's autopsy material.

MATERIAL AND METHODS

The study was based on the autopsy material from the Department of Pathmorphology, Medical Academy in Lublin collected in the period of 1984 – 1998. Of 6,157 autopsy protocols, 2,171 concerning children aged up to 1 year (body weight > 500 g) were chosen, regardless of live births or stillbirths.

Information about age, sex, place of residence or possible clinically diagnosed and surgically corrected congenital abnormalities came from the notes from the case history inserted in the autopsy protocol. Detailed data on congenital malformations that have been found in gross and/or microscopic examination came from the autopsy protocols, too.

We have used Mayer's definition of congenital malformation (4) i.e. deformation, anomaly or disturbances, that is present at birth, although in may not be clinically apparent at that time. Multiple congenital malformations were defined as involving at least two different systems, unless they presented known syndromes. In this case, they were included in the group of "others".

The occurrence and the structure of congenital malformations (sex and place of residence of children, kind of affected system) were estimated. Chi–square test with Yates' correction was used for the data analysis.

RESULTS

Congenital malformations were revealed in 903 cases (41.6%) of the 2,171 autopsy examinations in children up to 1 year of age. The highest incidence of congenital malformations was noted in 1985 and 1991 (89 and 81 cases, respectively), however the lowest incidence was seen in 1995 and 1998 (36 and 49 cases, respectively) (Table 1). The proportion of the autopsied children with developmental defects in Bach year of the analyzed period ranged from 29.0% to 53.3% (Table 1).

Congenital malformations occurred more frequently in boys (464 cases – 48.7%) than in girls (439 cases – 48.7%) and in children from the rural areas (500 cases – 55.4%) than in those from the urban areas (403 cases – 44.6%) (Table 1).

In the investigated population the most frequent were multiple congenital malformations (293 cases – 32.5%). The subsequent groups were defects of cardiovascular (217 cases – 24.0%), digestive (144 cases – 16.0%) and central nervous systems (134 cases – 14.8%). Abnormalities of other systems were much less frequent (Table 2).

We have found some sex and place of residence differences in the occurrence of congenital malformations of the particular systems. Namely, cardiovascular system, respiratory tract, genitourinary tract, digestive system and skeletal system malformations were frequently seen in boys, whereas central nervous system and multiple congenital malformations occurred frequently in girls. The differences concerning genitourinary tract and central nervous system malformations were statistically significant ($p< 0.01$ and 0.05, respectively) (Table 3). We have also found slightly higher incidences of genitourinary tract and skeletal system abnormalities among children from the urban areas. On the contrary, cardiovascular system, respiratory tract, digestive system, central nervous system and multiple congenital malformations were more frequent among children from the rural areas, but the differences did not reach statistic significance (Table 3).

We have also studied the structure of multiple congenital malformations. The most frequently seen elements of those abnormalities were malformations of digestive (52.9%), cardiovascular (44.4%) and skeletal systems (42.7%) (Fig. 1).

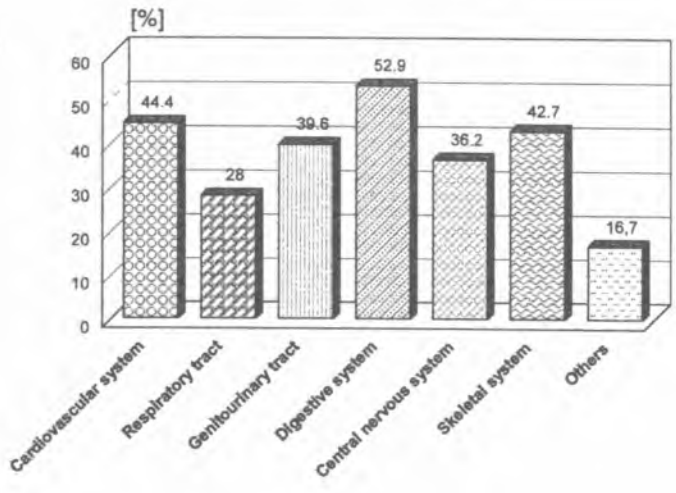

Fig. 1. The share of the defects of the particular systems among the multiple congenital malformations

DISCUSSION

The majority of the literature data concerning congenital malformations is based on the analysis of the clinical material (3, 4, 10, 11, 14), sometimes supplemented by the autopsy examinations (1, 6, 7, 8, 13). The studies based, like ours, on autopsy material only are rather rare (2, 5, 9, 15, 17), therefore the comparison of the results, and the expression of some general rules are difficult. It is obvious, that the analyzed autopsy material is selected, including cases in which congenital malformations were either the cause of death, or the factor of worsened prognosis and the death resulting from some other causes, or finally, they were only accidental autopsy findings, without any influences on the clinical course. It should be emphasized that the autopsy often reveals the presence of clinically not diagnosed congenital defects. On the other hand, many children with developmental defects survive thanks to modern method in therapy including surgical correction of the abnormality or absence of connections with children physical condition.

In our study, the percentage of autopsied children with congenital malformations was very high (41.6%), comparable only with data from Medical Academy in Białystok in the years 1986–1990 – 40.4% (17). Other investigators of autopsy material showed lower values (2, 9). As other authors (1, 10, 13, 14) we did not observe an increasing tendency in occurrence of developmental defects. Over the last 15 years the percentage of autopsied children with congenital defects maintained on the similar level.

We have confirmed the data of many investigators studying clinical (1, 3, 6, 8, 13, 14) as far as autopsy material (9, 17) about preponderance of boys in the group of malformed children (51.3%). It is probably connected with higher susceptibility of male embryos to teratogenic factors (1, 18).

In this report a higher occurrence of congenital defects among children from rural areas (55.4%) was found. The causes of this phenomenon are complex. They may be connected with hard physical

Tab. 1. The incidence of congenital malformations in children up to 1 year of age in the years 1984 – 1998 acc. to their sex and place of residence

Year	No. of children up to 1 year of age	No. of children up to 1 year of age with congenital malformations				
		total n (%)	boys n	girls n	urban areas n	rural areas n
1984	140	63 (45.0)	32	31	33	30
1985	167	89 (53.3)	49	40	36	53
1986	139	57 (41.0)	30	27	24	33
1987	140	62 (44.3)	39	23	29	33
1988	127	55 (43.3)	30	25	18	37
1989	147	57 (38.8)	37	20	28	29
1990	151	61 (40.4)	26	35	21	40
1991	152	81 (53.3)	38	43	42	39
1992	170	75 (44.1)	40	35	32	43
1993	160	51 (31.9)	21	30	24	27
1994	157	56 (35.7)	31	25	31	25
1995	124	36 (29.0)	13	23	13	23
1996	133	57 (42.9)	38	19	25	32
1997	133	54 (40.6)	21	33	24	30
1998	131	49 (37.4)	20	29	22	27
Total n (%)	2171 (100)	903 (41.6)	464 (51.3)	439 (48.7)	403 (44.6)	500 (55.4)

Tab. 2. The incidence of the congenital malformations of the particular systems in children up to 1 year of age in the years 1984-1998

Year	No. of children up to year of age with congenital malformations								
	Cardio-vascular system	Respiratory tract	Genito-urinary tract	Digestive system	Central nervous system	Skeletal system	Multiple congenital malformations	Others	Total
1984	14	0	3	11	16	0	18	1	63
1985	19	0	4	25	14	3	22	2	89
1986	17	1	2	8	11	1	15	2	57
1987	17	1	5	10	8	2	15	4	62
1988	17	2	2	12	9	0	11	2	55
1989	12	0	5	12	6	0	18	4	57
1990	12	0	0	12	9	2	23	3	61
1991	25	0	2	10	16	0	26	2	81
1992	18	0	2	8	6	2	34	5	75
1993	11	2	5	4	5	0	19	5	51
1994	12	0	0	10	5	0	22	7	56
1995	6	0	2	5	10	0	10	3	36
1996	13	0	2	8	5	5	21	3	57
1997	9	1	4	5	6	2	23	4	54
1998	15	0	2	4	8	0	16	4	49
Total n (%)	217 (24.0)	7 (0.8)	40 (4.4)	144 (16.0)	134 (14.8)	17 (1.9)	293 (32.5)	51 (5.6)	903 (100)

Tab. 3. The differences in the incidence of congenital malformations of the particular systems in children depending on sex and place of residence (NS – not statistically significant)

Congenital	Boys n	Girls n	p	Urban areas n	Rural areas n	p
Cardiovascular system	122	95	NS	95	122	NS
Respiratory system	6	1	NS	3	4	NS
Genitourinary tract	30	10	0.01	21	19	NS
Digestive system	84	60	NS	62	82	NS
Central nervous system	53	81	0.05	58	76	NS
Skeletal system	11	6	NS	9	8	NS
Multiple congenital malformations	136	157	NS	126	167	NS
Others	22	29	NS	29	22	NS
Total n (%)	464 (51.3)	439 (48.7)		403 (44.6)	500 (55.4)	

work of country women, their exposition to chemical agents and zoonosis (e.g. toxoplasmosis). We should not forget lower economic conditions, inappropriate food habits as far as contamination of ground water in rural areas (11, 18, 19). Nevertheless, the results presented in the literature are divergent. Some studies confirmed our observations (3, 11), wheras others indicated higher incidence of congenital defects in urban areas (5, 7, 8, 14).

We have found a higher proportion of multiple congenital malformations (32.4%) than cardiovascular (24.0%), digestive 16.0%) and central nervous systems (14.8%) abnormalities in the analysed population. The presented data were very similar to those of Sulkowski et al. (17), and the differences related only to central nervous system defects which in their study, came third ahead digestive system. According to many investigators, the most common malformations are cardiovascular ones (1, 4, 6, 8, 9, 13, 16). But some others thought that the skeletal system abnormalities are the most frequent (3, 10, 13). There is a domination of multiple congenital malformations in newborns born before term (1) and newborns with low birth weight (7). These differences probably result from the way of selection of the analysed population (clinical material vs. autopsy material). It is worth to emphasize that in our study cardiovascular defects were also noted in 130 cases within the group of multiple congenital malformations, so with regard to isolated malformations of this system there is confirmed domination of cardiovascular defects among congenital malformations.

In the present study, there were some sex differences in the occurrence of congenital malformations of the particular systems. Like in some other reports (3, 6, 15), central nervous system defects were significantly more frequent in girls, but those of genitourinary tract were more freguent in boys.

It is possible that biologically weaker mate embryos with severe brain anomalies had died early in utero, which led to spontaneous abortion (18).

The results of our study are highly comparable with the results of analysis in the years 1986–1990 from the Białystok group (17). The reasons are perhaps similar genetic material from the east part of Poland and/or influence of similar environmental teratogenic factors.

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STRESZCZENIE

Przeanalizowaliśmy częstość występowania oraz strukturę wad wrodzonych (płeć, miejsce zamieszkania, układ, którego dotyczyła wada) u dzieci do pierwszego roku życia w materiale autopsyjnym Katedry i Zakładu Patomorfologii AM w Lublinie z ostatnich 15 lat. Wady wrodzone występowały w 41,6% przypadków sekcji z tej grupy wiekowej. Nie stwierdziliśmy tendencji wzrostowej występowania wad w analizowanym okresie. Wady wrodzone były częstsze u chłopców oraz u dzieci z terenów wiejskich. Najliczniejsze były mnogie wady rozwojowe oraz wady układu sercowo–naczyniowego i pokarmowego. Stwierdziliśmy istotnie częstsze występowanie wad układu moczowo–płciowego u chłopców, a centralnego układu nerwowego u dziewcząt. Prezentowane wyniki wykazują dużą zbieżność z danymi uzyskanymi w ośrodku białostockim.