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Birth prevalence of congenital cardiovascular malformations in Hungary

by

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In a certain area of Szolnok county, 5,978 live births occurring in the period 1963—1965 were studied by the optimal epidemiological model. Of the study population, 5,433 children (91%), were identified. It was completed by 211 children, thus the total material involved 5,644 cases, and 67 congenital cardiovascular malformations were detected among them. Seven, being part of multiple congenital abnormalities, were excluded. Thus the birth prevalence of congenital cardiovascular malformations was 10.6 per 1000 live births.

In an earlier study the birth prevalence of congenital cardiovascular malformations (CCM) was found to amount 10.2 ± 2.1 per 1000 liveborns in 1963 in a defined region (Szolnok county) of Hungary [4]. A higher rate was observed only among Eskimos in Greenland [2]. The high Hungarian value was probably due to the effectiveness of the "optimal" epidemiologic approach. A single survey cannot, however, prove the validity of the high birth prevalence; the rate may have shown a particular value in children born in 1963 or in the surveyed area. Owing to this possibility our early sample was completed with the live births of 1964 and 1965.

MATERIALS AND METHOD

The live births of 1963—1965 in a certain area of Szolnok county residents were studied in two different approaches. The "Szolnok county population I" involved all the 3 290 babies born in 1963 to the inhabitants of Szolnok town and district as well as Törökszentmiklós town and district. Cardiological examinations were conducted in 1971—1972. "Szolnok county population II" included all the 3 682 babies born in the years 1964 and 1965 to the inhabitants of Szolnok town and district. Cardiological screening and the evaluation of these cases were made in 1974—1978. Owing to the similarity of epidemiological features in these two populations and to use of the same method, these cases will be evaluated together.

We received from the Central Statistical Office the list of the names of the live borns

in the years of 1963—1965 in Szolnok county. We planned personal examinations by the same paediatric cardiologist in every liveborn of this population involving 5 978 cases. (Stillborn babies had to be ignored, because most of them were not subjected to post mortem in that county.) In this population, known by name in advance for every child, the case ascertainment proceeded as follows.

(a) The children born in 1963—1965 attended primary school at the time of the study, i.e. in 1971—1978. Thus, the paediatric cardiologist visited the 89 primary schools in the area and examined 4 906 children on the list (82.1%). She also examined 211 children born in 1963—1965 outside of Szolnok county.

(b) The data for 233 children born in 1963—1965 in the region examined but deceased up to the time of study (3.9%) were evaluated on the basis of the files of the pathology department and the forensic medical records. Of the infants who died 92%, and of the children who died about 80%, were subjected to necropsy. In this way we obtained relevant information on the fatal cases of CCM.

(c) A small proportion of the children domiciled in the region studied had been excused from going to school because of mental subnormality or for other reasons. The names of these 294 children (4.9%) were obtained from the Szolnok County Council. 164 children cared for in various institutions were visited and a cardiological investigation was performed. It was beyond our resources to visit the 130 children who were being cared for in their homes. Medical information on them was requested from paediatricians by the help of the parents.

(d) From the Szolnok County Council we obtained the list of the 481 children who had moved from the county in the period 1963—1978, with their new adresses. The children on the address list, 8.0% of the study population, were living in different parts of the country, therefore we have been unable to reach them.

According to our hypothesis, we should have found every child on the list of names. Still, 64 cases (1.1%) had "disappeared". Some of these children might have been taken for treatment to medical institutions outside Szolnok county where they may have died or were autopsied, and technical errors too may have occurred when checking and counter-checking the lists.

To replace the persons who moved, we used the 211 children mentioned under (a) born in 1963—1965 outside Szolnok county, who had been subjected to cardiological examination in school. They reprented 3.5% of the whole sample. Just like in the case of those who had moved, in their case selective effects relative to CCM did not have to be taken into consideration.

The uniformity and reliability of the cardiological examination were ensured by a senior paediatric cardiologist [4]. Those cases which had been suspect of having some heart anomaly were asked to report, accompanied by their parents, at the Paediatric Cardiology Clinic, Szolnok, where detailed cardiological examinations were then performed.

Results and Discussion

In the so-called "optimal" epidemiological survey, 5 433 children (90.9%) of the Szolnok county population were indentified. It was completed by 211 children born outside of Szolnok county in 1963—1965 but attending the primary schools examined. Thus, in the total material of 5.644 (94.4%), 36 cases of CCM were detected by cardiological screening. Studies of the necropsy records disclosed further 31 verified cases of CCM. These 67 cases (Table I) indicate essentially a total birth prevalence

TABLE I

Distribution of persons in the study population

Groups and subgroups		Szolnok and Törökszentmiklós town and district 1963	Szolnok town and district 1964	Szolnok town and district 1965	Total (1963—1965)
Number of live births (Central Statistical Office)		2,296	1,908	1,774	5,978
Total evaluated cases		2,259	1,742	1,643	5,644
Total (unequivocal and doubtful) CCM cases		29	24	21	74
Total (unequivocal) CCM cases		23	23	21	67
Isolated CCM cases		17	14	12	43
Complex CCM cases		5	6	6	17
Together	No.	22	20	18	60
	Per 1000	9.7	11.5	11.0	10.6
Multiple CCM cases		1	3	3	7

of CCM of 11.9 per 1000. Seven CCMs were, however, part of multiple congenital abnormalities, thus the birth prevalence of CCM was 10.6 per 1000 live births.

CCM could seriously be suspected in 7 more cases. Two children are under observation because of stenosis of the aorta. In 4 newborns CCM was diagnosed clinically, but they died and no necropsy was performed on them. (One of these CCMs was certainly part of a multiple congenital abnormality.) One index patient has Wolff—Parkinson—White syndrome. If we accepted these 6 isolated or complex CCM cases of doubtful diagnosis, too, the birth prevalence would increase to 11.7 per 1000. With the multiple CCMs the total birth prevalence would be 13.1 per 1000.

The observed distribution of CCMs is shown in Table II. The birth prevalence of isolated types of ventricular septal defect, aortic stenosis, patent ductus arteriosus, pulmonary stenosis and atrial septal defect type II were 1.9, 1.4, 1.2, 1.1 and 0.9 per 1000 live births, respectively. Of the 17 complex CCMs 2 each were endocardial fibroelastosis and transposition of great vessels. Of the remaining 13 complex CCMs, 5 each had ventricular septal defect and patent ductus arteriosus. Of these, only two did not die in infant age. Only one case (Kartagener syndrome) of multiple CCMs is alive at the time of evaluation.

The 10.6 per 1000 birth prevalence of CCM includes only isolated and complex CCMs in the liveborn babies and the cases with verified diag-

TABLE II

Distribution of categories and types of congenital cardiovascular malformations (CCM) in the study population (The number of deaths is shown in brackets)

Isolated CCM \ Year	1963	1964	1965	Total No	Total ‰
VSD	4 (1)	3	4 (1)	11	1.9
PDA	3	4 (1)	0	7	1.2
ASD I	1	0	0	1	0.2
ASD II	3	1 (1)	1 (1)	5	0.9
PS	1	3	2	6	1.1
AS	2 (1)	2	4	8	1.4
CA	2	1	0	3	0.5
Dextrocardia	0	0	1	1	0.2
Ebstein	1	0	0	1	0.2
Total	17 (2) 7.5‰	14 (2) 8.0‰	12 (2) 7.3‰	43	7.6

Complex CCM

1963		1964		1965	
TC	1 (1)	VSD + PDA + TGV	1 (1)	VSD + PS	1
VSD + PDA + AS	1 (1)	FT	1 (1)	VSD + PS + Valv	1 (1)
VSD + ASD + Valv	1 (1)	ASD II + PDA	2 (1)	ASD + TC	2 (2)
TGV	2 (2)	PDA + CA	1 (1)	PS + Valv	1 (1)
		EF	1 (1)	EF	1 (1)
Total	5 (5) 2.2‰		6 (5) 3.4‰		6 (5) 3.7‰
1963—1965		17 3.0‰			

Multiple CCM

1963		1964		1965	
VACTERL (CCM)	1 (1)	Down (ASD I)	2 (2)	Down + PDA	1 (1)
		Situs inversus (Dextro, VSD + PDA + + PS)	1 (1)	Holt-Oram (VSD + ASD)	1 (1)
				Kartagener (Dextro)	1
1963—1965		7 1.2‰			

Abbreviations:
VSD = Ventricular septal defect
PDA = Patent ductus arteriosus
ASD = Atrial septal defect
PS = Pulmonary stenosis
AS = Aortic stenosis
CA = Coarctation of aorta
TC = Truncus communis
TGV = Transposition of great vessels
Valv = Valvular anomaly
Dextro = Dextrocardia
EF = Endocardial fibroelastosis

nosis. Three important lessons could be derived in the course of evaluation of this high CCM prevalence: (i) The true birth prevalence of CCMs may be around 10 per 1000 [6]. This, however, can be brought to light exclusively by an adequate, optimal epidemiological approach ascertaining all CCM cases. (ii) The birth prevalence found in the Szolnok county population may be a result of a regional clustering [3]. The possibility may be corroborated or rejected on the basis of studies by this method extended to other territories. (iii) Paediatric cardiological screening is of great practical value: in 18 of the 36 cases diagnosed in vivo, CCM was detected as a result of screening. Prior to that, neither the parents nor the teachers had had any knowledge of the condition which, of course, was not severe.

The birth prevalence of CCMs published in the literature varies from 0.1 per 1000 to 18.5 per 1000 [4]. The 185-fold frequency spectrum is unrealistic, and can be explained by the difficulties of such studies in the first place. Some of the reasons which may be involved are, (i) differences in the definition of CCM; (ii) differences in the populations studied: stillbirths and/or liverbirths, postmortem and/or clinical material, selected material, differences in age; (iii) differences in diagnostic methods (pathological, special surgical, general clinical, cardiological); (iv) differences in the ascertainment; (v) differences in conceptual approach, in preparedness and consciousness; (vi) differences due to variations in the size of the sample populations; and, finally (vii) the duration of follow-up, in view of the fact that certain CCMs become manifest late.

An improvement in the technical standard of epidemiological survey is indicated by the fact that while in reliable surveys of the 'fifties 3 to 4 per 1000, in the 'sixties 6 to 8 per 1000 values were obtained for CCM occurrence [1]. The real birth prevalence is, however, expected to amount to 10 per 1000. Thus CCM, as a whole, constitutes one of the most common organ localisations of congenital abnormalities.

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