THE RETROSPECTIVE STUDY ON THE ETHIO-PATHOGENIC FACTORS OF THE CONGENITAL MALFORMATIONS

- in the Bihor district-

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The congenital malformations represent a major cause of mortality and morbidity in childhood; to prevent this, it is important to know the level of morbidity and its etiology.

Knowing the level of the diseases prevalence as well as the ethiopathogenetic factors is necessary to obtain a real image of the congenital malformation phenomenon, image which will have the role of creating the strategies and measures to be used in this field.

This study was designed to provide scientific evidence in order to formulate hypothesis regarding the strategical approach of the problem caused by the congenital malformations among of the underage population from the Bihor district as well as to apreciate the impact which the congenital malformations have upon the people's health.

Key words: congenital anomalies, ethiopathogenetic factors, health promotion.

Introduction

The congenital malformations represent a current problem in human pathology as a result of frequency and ethio-pathogenic aspects as well as of the socio-medical implication.

Numerous international, na-tional and regional statistics estimate that the prevalence of the congenital malformations, detectable by birth and clinical examination of new born living and dead, ranges between 1.5 – 2% [3] (COVIC, 1981).

In the Bihor district there is a specialized service, the genetic department of the Municipal Hospital „Gavril Curteanu" from Oradea, whose activity is to detect malformations defects in newborn departments and pediatric hospitals in the Bihor district.

The specific activities of this department are:

- Early detection and monitoring of the genetic diseases and congenital abnormalities in the investigated territory;
- Better knowledge of the structural and ethiological aspects of the congenital anomalies;
- Knowledge of the frequency and other epidemiological information about genetic diseases and congenital anomalies in the observed territory;
- Identification of the role and the place of the congenital abnormalities in the public health and the possibility of reducing their impact on family and society.

Given the experience of 22 years of activity in this specialized field for diagnosis, hospitalization, recovering and genetic counceling, we consider that our point of view regarding to both the benefits disapointments as well as the results obtained in this department can be considered as references in identifying the necessities and obstacles encountered by the team studying the problem of congenital abnormalities in Romania.

The aim of the present study was to analyse, from the epidemio-logic point of view, the situation of the congenital malformation among the children in Bihor district.

The main objectives of the present study were to identify:

- The number of children born with congenital malformations in the studied period (1984 – 2000);
- The degree of their health status affectation according to the number of diagnosis;
- The type of existing malformation at these children;
- The frequency of the ethiopathogenic factors incriminated in the congenital pathology malformations.

In order to fulfil these ***objecti-ves***, the following activities were plann-ed and achieved:

- The registration of the number of alive new born during 1984 – 2005 in the Bihor district;
- The analyse of the morbidity of the congenital abnormalities (the incidence of the cases registred in this period in the Genetic Department of the Municipal Hospital „Gavril Curteanu");
- The patients' classification according to the number of diagnoses per patient;

- The identification of the ethiopathogenetic factors of the congenital malformation discovered in the studied group;
- The frequency analysis of the congenital abnormalities according to pathogenetic mechanism;
- The territorial distribution description of the congenital malformations in the Bihor district;
- The distribution of the congenital abnormalities by residence location, the mother's level of education, parent's ages in the moment of conceiving, the gender of children with congenital abnormalities.

Material and methods

The present study is a transversal and retrospective analyse based on the existed data collected from the Genetic Departments of the Municipal Hospital „Gavril Curteanu" from Oradea, during a period of 22 years (1984 – 2005).

The criteria of including the cases in this study: this research includes all the patients from the Bihor district with congenital ab-normalities who were selectively monitored by our department in the period of study.

The criteria of exclusion of the cases from the study: there were excluded from this study all the new born babies declared dead by a specialist physician.

Detecting the congenital abnormalities were performed consecutively to a selection of the cases from all departments of new born babies in pediatric hospitals, ambulatory services, centres of children hosting from all the Bihor districtv and by the GPs and by pediatric specialist physician all over the district.

The continuous and active monitoring of the discovered cases involved in early solving and/or prevention of exacerbation, complications, cronicizations, invalidities, by a large interdisciplinary colaboration (surgery, pediatric orthopedy, oftalmology, ginecology, dermathology, endocrinology)

The source of data: it was represented by the medical records for each child monitored by the genetic service.

Each ***„record file" contains the following information:***

- patient's data;
- family history, the pregnancy and birth giving history;
- phisical examination and somatometrical measures;
- the result of the paraclinic and genetic investigations, imagistic procedures;
- diagnosis, genetic councelling;
- medical monitoring and recovery.

The patients were taken out of this active evidence in the following situations:

- When they are treated (complet recovery);
- In case of a change of the residence place;
- in case of death.

The cases which come out of the active evidence are placed in a passive evidence, separately, in which all the information from the active evidence are taken out.

Also, for each new case taken into evidence, a record of evidence is taken over and then the main data from this record are introduced into a pro-gramme of statistics processing.

Results

Between 1984 – 2005, in the Bihor district there were recorded approximately 169250 alive newborns according to the public data from the statistic of the Bihor Public Health Department. In the same time there were recorded 3377 children with congenital abnormalities representing 2% from the total number of the alive newborns in the studied period. The first phase in analysing the gathered data was represented by analysing the regional distribution of the congenital abnormalities. The 6 distinct territories differ by characteristics as well as the population number, natality living condition, morbidity and cultural model existing in region. Thus, in territorial context, there have been observed high levels of prevalence especialy in Oradea (2.377 cases); also, high prevalence was recorded in region such as: Marghita (469 cases), Beiuş (293 cases), Aleşd (268 cases), Salonta (205 cases) and low prevalence in Ştei (116 cases).

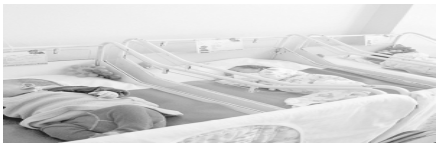
The distribution of congenital abnormalities by residence reveals a clear preponderence of the cases in the rural regions in comparison with the urban regions.

The mothers' age. Among children with congenital abnormalities, 29.71% had mother who was not yet 20 years old, and 9.04% had mother being over 35 years old. The importance of the level of education for the mothers with children having congenital abnormalities is moti-vated by the fact that 21.1% had a low level of instruction, respectively 3.05% were illiterated and 18.05% had only finished the primary school. The education combined with age and mother's residence (which predominantly was in rural) have a major influence both on the way in perception and addresing for a prenatal visit to a physician and on the possi-bility of finding out the first signs of disseases and then addressing to medi-cal services to get a proper medical assistance.

The number of CA per case

As the number of congenital abnormalities detected was bigger than the number of the children with congenital abnormalities, in this study there was taken into account an indicator which quantifies the number of congenital abnormalities per case.

So, there were identified 2.849 (84.36%) cases with unique diagnosis of congenital abnormalities, 307 (9.09%) with two diagnosis, 123 (3.65%) with three diagnosis and 98 (2.90 %) with more than three diagnoses of congenital abnormalities. The patients with heart conge-nital malformations were considered with only one diagnosis.



The type of congenital abnormalities. Regarding the type of congenital abnor-malities, there were iden-tified 218 entities in this study; the malformations, dysplasia and deformations are the most frequent enti-ties being encountered (table 1).

Approximately 50% of the diagnosed congenital abnormalities were represented by malformations and only 10% were represented by the dysplasia (11.7%), respectively by the deformations (9,.%).

Table 1. The distribution of cases by type of CA, Bihor district, 1984-2005

Clinical-genetic variants (entities)	Solitaire		Asociated		TOTAL	
	No.	%	No.	%	No.	%
Malformations (46 entities)	*1339*	*40.4*	*641*	*64*	1980	**45,9**
Disruptions (9 entities)	*76*	*2.3*	*27*	*2.7*	103	**2,4**
Deformations (6 entities)	*349*	*10.5*	*59*	*5.9*	408	**9,5**
Dysplasia (24 entities)	*413*	*12.5*	*91*	*9.1*	504	**11,7**
Other unclasified AC (20 entities)	*166*	*5.0*	*98*	*9.8*	264	**6,1**
Specificated multiple AC (113 entities)	*968*	*29.2*	*84*	*8.4*	1052	**24,4**
TOTAL (218 entities)	**3311**	**100**	**1000**	**100**	**4311**	**100**

Data source : *Genetic Department of the Municipal Hospital „Gavril Curteanu" from Oradea,* Electronic evidence of the cases with congenital abnormalities, 1984-2005

Table 2. The distribution by CA groups according to the OMS classiffication, Bihor district, 1984-2005

The congenital abnormalities (CA) group	No of cases with diagnostic	% of total cases	Prevalence per 100.000 new borns
CA localizated at the osteo-articular system	798	18.5	4.16
CA of the circulatory system	719	16.7	2.18
Benign and malignant tumors	417	9.7	3.75
CROMOZOMOPHATIES	280	6.5	1.46
CA of the nervous system	270	6.3	1.41
CA of genital organs	211	4.9	1.1
CA of Oral-cavity	185	4.3	0.96
Illnesses of the digestive system	174	4	0.91
Other unclasified CA in other places of the body	171	3.9	0.89
Congenital malformations of the skin, facomatosis	142	3.3	0.74
Specificated syndroms, Teratogenics	138	3.2	0.72
Eyes illnesses (ophthalmic system illnesses)	109	2.5	0.57
CA of the urinary system	106	2.5	0.55
CA of the respiratory system and Otho-rhino-laryngology	103	2.4	0.54
Metabolism errors	102	2.4	0.53
Generalized MC of the osteo-articular system	101	2.3	0.51
Muscular diseases, Hernia	98	2.3	0.51
Central Nervous System, Neuromuscular Diseases, Mental Retard	84	1.9	0.44
Endocrin Diseases	39	0.9	0.2
Blood and immune system deficiency	35	0.8	0.18
Congenital infections	29	0.7	0.15
TOTAL	4311	100%	22.48

Data source : *Genetic Department of the Municipal Hospital „Gavril Curteanu" from Oradea,* Electronic evidence of the cases with congenital abnormalities, 1984-2005

The prevalence of the congenital abnormalities between 1984 – 2005, in the Bihor district (table 2)

To get evidence which can be compared to an international level, the prevalence of the congenital abnormalities can be calculated for the studied period according to the WHO clasification (sharing by functional human body systems).

Thus, In the studied period, the highest prevalence of the abnormalities was recorded for the osteo-articular system (4.16 at 100000 newborns), followed by abnormalities of circulatory system and respectively benign and malign cancers.

The chromozomopathies (280 diagnostics) represent 6.5% from the total identified congenital abnormalities; it was recorded a prevalence of 1.46 at 100.000 new borns for the studied period.

The ethio-pathogenic factors involved in the congenital abnormalities appearance

The genetic causes represent the most important factor in producing the congenital abnormalities in childhood.

Among the 4311 studied conge-nital abnormalities, the genetic factor was identified in a great number of cases; so, the genetic factor is recog-nized as a major cause of this illness (2953 cases representing 68.5%); the prevalence of the ethiopathogenic factors is ilustrated in table 3.

Table 3. Congenital anomalies distribution by ethio-pathogenic factors, in Bihor district, 1984-2005

ETHIOPATHOGENIC FACTOR	The no. of CA	% of total no. of CA
Genetic factors	2953	68.49
Chromosomal factors	289	6.7
Monogenic factors	728	16.9
Multifactorial – polygenic factors	1936	44.9
Theratogenic factors	38	0.89
Unknown factors	1320	30.62
TOTAL	4311	100%

Data source: *The Genetic Department of the Municipal Hospital „Gavril Curteanu" of Oradea; The electronic evidence of the cases with CA, 1984-2005*

Conclusions

During 1984 – 2005 period, there were recorded in the Bihor district approximately 169250 alive new borns and 4.311 identified congenital abnormalities in 3377 children. The diagnosis of unique congenital abnormality prevails and 2849 diagnoses respectively 84.36% from the total number of CA were recorded; the prevalence level was of 168.33 at 100.000 alive new borns.

Regarding the type of congenital abnormalities, in the present study there were identified 218 entities: 46 types of malformations, 9 types of disruptions, 6 types of deformations, 24 of dysplasia, 20 unclasified congenital abnormalities and 113 congenital abnormalities multiple specified.

According to the WHO classification, the biggest frequencies of congenital abnormalities were recorded for the osteo-articulary (18.5%) and circulatory system (16.7%).

From the ethiopathogenetic point of view (68.49%), we can apreciate that more than two thirds from the total of congenital abnor-malities presented genetic causes and only 0.89% presented theratogenic factors; the rest of the congenital abnormalities (30.6%) had as cause an unknown factor.

From the pathogenetic me-chanism point of view, the most frequent CA were:

- Dominant autozomal: neurofibromatosis, osteogenesis imperfecta;
- Recessiv autozomal: phenylketon-uria, albinism;
- X recessiv: Duchenne muscular dys-trophy, type A of hemophilia;
- Deformations: congenital crooked foot;
- Cromosomophaties: Down syndrome;
- Multifactorial polygenic: MCC;
- Theratogens: congenital toxoplasmosis;
- Disruptions: amniotic disease;
- Dyspplasia: hemangioma.

The congenital malformations are structural or functional defects presented at birth; they are determined by genetic factors or extern factors (terathogens), and by knowing them one can lead to the identification and quantification of the risk even before the child's conceiving.

The congenital malformation profilaxy must include integrated ac-tions sustained at all levels: primordial, primary, secondary and tertiary.

The approaches must be considered for the congenital malforma-tion profilaxy must be associated with complex educational actions for health addressed both to pregnant women and to population in general. They must be awared on risk factors, problem understanding, importance of the prenatal diagnosis as well as following the doctor's indications.

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