THE EPIDEMIOLOGICAL STUDY OF THE CONGENITAL MALFORMATIONS IN THE COUNTY OF BIHOR

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Abstract: *The congenital malformations are widespread all over the world, without latitude and longitude, with only regional differences regarding the clinical manifestations, etiology and pathology. The study formulates hypotheses regarding the incidence, prevalence and mortality through congenital malformations in the population of the county of Bihor.*

Keywords: *malformation, etiology, diagnosis, prevention*

Rezumat: *Malformaţiile congenitale sunt răspândite pe întreg globul, neexistând latitudine şi longitudine în care ele să nu fie prezente, existând doar diferenţe zonale în ceea ce priveşte manifestările clinice, etiologia şi patologia acestora. Studiul formulează ipoteze privind incidenţa, prevalenţa şi mortalitatea prin malformaţii congenitale la populaţia judeţului Bihor .*

Cuvinte cheie: *malformaţii, etiologie, diagnostic, prevenire*

INTRODUCTION

The congenital malformations frequency arrived in 2006 at 5% from the total of the births in comparison with only 4% in 2000. The estimation belongs to the Health Statistics Centre from the Ministry of Health.

Out of 200 000 of new-born children every year, more than 10 000 have malformations. Analyzing the causes of the perinatal mortality, 2/3 are generated by congenital malformations, diseases and sufferings from the perinatal period and only 1/3 by birth.(1)

The most frequent congenital malformation among the 20 000 is the Down syndrome with an incidence of 1: 800 births. This illness is characterized by psychic and physical retardation, having heart malformations in 30 – 40% of the cases.

The Down syndrome appears more frequently in the cases in which mothers are over 35 years old. Life expectancy of a person with Down syndrome is between 45 and 55 years old.

Another type of congenital malformation is the Edward syndrome, with an incidence of 1:8000 births, characterized through low weight at birth, short nose, small breast and serious mental retardation.(2,4)

OBJECTIVES

The aim of the study is to identify the number of new-born children with congenital malformations in this period, the type of malformation at these children, the incriminated causes in the congenital malformations pathogeny, paraclinical examinations used in their diagnosis, the frequency of the genetic advice and the mortality through congenital anomalies.

This study aims to bring a contribution to the:

- A better knowing of the present situation, of the dynamics and the structure of the specific mortality through AC;
- Its identification among population, of those category of persons, who in connection with congenital illness can present a priority in applying the prophylactic measures and education for health, this group being an important potential in reducing the morbidity and mortality;
- The systematization of some data and information referring to the specific morbidity and mortality through AC, the establishment of some bases of references in this field.

MATERIAL AND METHOD

The study of spreading and medical implications of the congenital malformations, of their evolution in time, represents the most adequate way of establishing some efficient measures in preventing these illnesses.

The method used in this study is observation, which consists in following the development of some phenomena to be able to analyse them in dynamics.

The present work is an observational study, retrospective and descriptive, developed on the basis of the data gathered and existent at the Genetic Department of the Municipal Hospital „Gavril Curteanu „ on a period between 1984 – 2008 and on the basis of some statistic materials of the Authority of Public Health Bihor and of the County Statistics Authority Bihor.

RESULTS

Between 1984 – 2008, in Bihor county, there were registered **189150 alive new-born children**, data come from the statistic materials of the Authority of Public Health Bihor. In the same interval of time, at the genetic department of the hospital, there were taken in evidence 3613 children with AC, these representing 2% from the total number of the live new-born children in the studied period.

Table no. 1. The classification of the patients with congenital anomalies according with the etiology

	Number of AC
Genetic causes	**1736**
Chromosomal	**285**
Monogenic	**357**
Poligenic-multifactors	**1094**
Theratogenic factors	**46**
Unknown factors	**1798**
TOTAL	**3580**

According to the pathogenic mechanism and causes, the congenital anomalies more frequent in this period are:

- Out of 196 cases with dominant autosomal transmission – 30,6% neurofibromatosis, 10,7% polycystic infantile kidney disease, 10,7% osteogenesis imperfecta;
- Out of 145 cases with recessive autosomal transmission – 17,9% ocular - cutaneous albinism and 17,9% phenylketonuria;
- Out of cases with X linked-recessive transmission – 48,1% Duchenne muscular dystrophy ;
- Out of chromosomophaties, an important percent of 90,35% represents the Down syndrome;
- The heart congenital malformations represent 50,5% from the malformations with polygenic transmission;
- Out of the malformations determined under the teratogenic factors, 66,66% were obvious congenital toxoplasmosis

Table no. 2. Methods of diagnosing and preventing the congenital malformations, used in the period 1984 – 2008

AC according to etiology	Methods of diagnosing	Genetic advice	Mortality
Chromosomal causes	Caryotype – 36 determinations Echography – 21 X - ray - 14	25 consultations	84 deaths
Monogenic causes	Caryotype – 3 Echography – 10 X - ray - 26	37 consultations	48 deaths
Polygenic causes	Caryotype – 3 Echography – 14 X - ray - 30	30 consultations	45 deaths
Teratogenic causes	Caryotype – 1 Echography – 5 X – ray - 4	3 consultations	14 deaths

CONCLUSIONS

1. Between 1984 – 2008 there were recorded in Bihor county 189150 alive new-born children and 3613 children with congenital anomalies;
2. From the ethiopathogenetic point of view, 48,49% from the total AC had genetic causes and only 1,2% theratogenic factors;
3. From the pathogenetic mechanism point of view, the most frequent AC were: neurofibromatosis 30,6%, osteogenesis imperfecta 10,7%, phenylketonuria, albinism 17,9%, Duchenne muscular dystrophy 48,1%, Down syndrome 90,35%, cardio-circulatory malformations 50,5%, congenital toxoplasmosis 66,66%;
4. The diagnosis methods used more frequently were: caryotype determination, echography, X - ray, CT, RMN, hormones and enzymes dosing, ADN, hematology and biochemical tests, dermatoglife and Barr test, immunology;
5. There was given genetic advice for 25 patients with AC with chromosomal causes, 37 with monogenic causes, 30 with polygenic causes and 3 with teratogenic factors;
6. The mortality showed a growing among patients with AC with chromosomal causes (84 cases);
7. For an efficient prophylaxis of the genetic diseases, it is necessary satisfying of at least 3 conditions: the genetic education of doctors and other medical stuff; training for specialists and developing of an adequate structure for consulting, diagnosing and prophylaxis of the genetic diseases; starting some actions or regional and national programs for preventing the genetic diseases.(3)

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