STUDY ABOUT THE INCIDENCE OF GENETIC MALADIES AND CONGENITAL MALFORMATIONS IN HUMAN POPULATIONS FROM BACĂU COUNTY IN 2006

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Key words: genetic diseases, congenital malformation, prophylactic

Abstract

Genetic diseases are very numerous in humans. Currently, we know about 10.000 diseases that are determined or conditioned by genetic factors. These maladies have a great diversity and affect all kinds of systems and organs therefore we could find them in every medical area.

In this paperwork I have studied the frequency of genetic maladies and congenital malformation during the year 2006 for the children in Bacau County. This initiative came to support the „National Programme for prophylactic methods in genetic disorders".

The results show that male children are the most affected by congenital malformations. The samples were represented by a group of male children (12 subjects) compared to a sample of 8 female subjects. We have chosen this number of subjects because it is representative for the number of male and female births during the year 2006.

Introduction

Genetic diseases are chronicle diseases that frequently cause physical, sensory, motor or mental disabilities. They are the cause for 75% of mentally retardation, deafness or blindness in children. The chronicle nature of many genetic diseases imply o great medical, financial and emotional burden for the patients and their families, as well as for society in general

Thus, genetic diseases represent o major public health issue that implies concrete and constructive actions towards a good diagnose and efficient prophylaxis. In this paper I have studied the frequencies of genetic maladies and congenital malformations in the year 2002 amongst the children form Bacău city in order to support *The National Program for Prophylaxis of Genetic Diseases*.

Presentation of the cases studied in 2006

In 2006 I have studied 4413 subjects who were patients of The County Pediatric Hospital. Amongst these 27 children were diagnosed with diverse genetic maladies and congenital malformations.

The 27 cases (see Table 1) are distributed as it follows:

- 3 cases of *hemolytic anemia*;
- 4 cases of *mental retardation*;
- 20 cases of *congenital malformations*.

Table 1. Distribution and frequency

Effect	Nr. of subjects	Distribution and frequency (%) of the cases	
Hemolytic anemia	4413	3	0,068
Congenital malformations	4413	20	0,453
Mental retardation	4413	4	0,09
Total	4413	27	0,612

From the 27 cases that we took under observation we have registered 6 deaths through the year of 2006 from the ones with congenital malformations (see table 2).

Table 2. Distribution and frequency of deaths registered in 2006

Effect	No. of subjects	Distribution and frequency (%) of deaths	
Hemolytic anemia	3	-	-
Congenital malformations	20	3	15
Mental retardation	4	-	-
Total	27	3	11,11

Congenital malformations are represented by a number of 27 cases that we took under observation, 18 boys and 9 girls (table 3). The diagnostics were confirmed by the physicians from The County Pediatric Hospital and other specialized clinics that the patients consulted afterwards. The cases have prenatal debut and could have been confirmed through ultrasound investigations in the last months of pregnancy and were confirmed after a specialized consult and clinical investigations. According to a study of medical statistics that took place in 2004 and was brought up by the Ministry of Health, at least 4-5 % of the children that are born in Romania present a malformation of some type, 6% being kids over the age of two and 8% are children over the age of 8.

From the total number of cases that we studied, we have found three bays with hemophilic anemia and three boys with mental retardation (table 3), cases that were confirmed by laboratory explorations.

The age group that had the highest number of cases (we are talking about the age when we found the disease, because some diseases may appear at birth, but they are later diagnosed, usually at a general check up, vaccination or a childhood disease) is the group between *0-1year with* 18 cases, followed by the group between 5-7 years (kindergarten and school) with 4 cases, the age group of over 10 years with 3 cases and the groups *3-5 years* and *7-10 years*, each with one case (see table 4).

Table 3. Sex distribution and frequency of the cases

Effect	No. of cases	Distribution and frequency (%) of the cases			
		Girls		Boys	
Hemolytic anemia	3	-	-	3	100
Congenital malformations	20	8	40	12	60
Mental retardation	4	1	25	3	75
Total	27	9	33,33	18	66,67

Tabelul nr. 4. Age group distribution and frequency of the cases

Effect	No. of cases	Distribution and frequency (%) of the cases				
		0-1	3-5	5-7	7-10	over 10
Hemolytic anemia	3	1(33,34)	-	-	-	2(66,67)
Congenital malformations	20	15(75)	-	3(15)	1(5)	1(5)
Mental retardation	4	2(50)	1(25)	1(25)	-	-
Total	27	18(66,67)	1(3,7)	4(14,82)	1(3,7)	3(11,11)

The population was preponderantly rural – 14 cases, followed by those who came from Bacau city – 10 cases and other cities – 3 cases (see table 5). The rural environment „suffers" from lack of specialized medical personnel, and the medical facilities are very poorly endowed with medical equipment, beneath the minimal standards.

Tabelul nr. 5. Rural and urban distribution and frequency

Effect	No. of cases	Number and frequency (%) of the cases		
		Urban area		Rural area
		Bacău	Other cities	
Hemolytic anemia	3	-	-	3(100)
Congenital malformations	20	7(35)	3(15)	10(50)
Mental retardation	4	3(75)	-	1(25)
Total	27	10(37,04)	3(11,11)	14(51,85)

Congenital malformations represent the majority of the investigated cases, thus being the object of our study, as there are 20 cases of this type. In this group we include:

- *Congenital malformations* of the heart – 9 cases ;
- *Hydrocephaly* – 3 cases;
- *Congenital malformations of the renal system* – 3 cases;
- *Congenital malformations of the fingers and toes* – 2 cases;
- *Microcephaly, Congenital Mega colon* and *Multimalformation syndrome* – one case each (see table 6).

Tabelul nr. 6. Congenital malformation distribution and frequency

Congenital malformations	**NO. OF CASES**	Distribution and frequency (%) of the case	
Congenital malformations of the heart	20	9	45
Hydrocephaly	20	3	15
Microcephaly	20	1	5
Congenital malformations of the renal system	20	3	15
Congenital malformations of the fingers and toes	20	2	10
Congenital Mega colon	20	1	5
Multimalformation syndrome	20	1	5

A major risk factor in malformations is the low age of the mothers, teenagers, who are at their first pregnancy. We consider the age group „12-15 years" and „16-18 years", the risk factor at this age groups being very high, – 5% for the first category and 1% for the second.

In teenage mothers we often find factors like: risky sexual behavior, unintentional pregnancy, reduced birth weight, premature birth, induced traumatism at birth, prolonged birth period, anemia, toxemia, caesarian section, uterine birth retardation, congenital malformation (1).

Table 7. Congenital malformation distribution and frequency by sexes

Congenital malformations	**NO. OF CASES**	Distribution and frequency (%) of the case			
		Girls		Boys	
Congenital malformations of the heart	9	3	33,33	6	66,67
Hydrocephaly	3	2	66,67	1	33,33
Microcephaly	1	1	100	-	-
Congenital malformations of the renal system	3	1	33,33	2	66,67
Congenital malformations of the fingers and toes	2	-	-	2	100
Congenital Mega colon	1	-	-	1	100
Multimalformation syndrome	1	1	100	-	-
Total	20	8	40	12	60

Boys are the most affected by congenital malformations, being represented by a sample of 12, in comparison with the girls that are represented by a sample of 8 cases (table 7).

The age group *0-1year* presents 15 cases, confirmed and reported, followed by the 5-7 years age group cu three cases, and the over 10 years age group and 7-10 age group with one case each. The 1-3 years and 3-5 years age groups do not have any representatives (see table 8).

Table 8. Age groups distribution and frequency of congenital malformation cases

Congenital malformations	Number of cases	Age groups and frequency (%)			
		0-1 year	5-7 years	7-10 years	over 10 years
Congenital malformations of the heart	9	7(77,78)	1(11,11)	-	1(11,11)
Hydrocephaly	3	3(100)	-	-	-
Microcephaly	1	1(100)	-	-	-
Congenital malformations of the renal system	3	1(33,33)	1(33,33)	1(33,33)	-
Congenital malformations of the fingers and toes	2	1(50)	1(50)	-	-
Congenital mega colon	1	1(100)	-	-	-
Multimalformation syndrome	1	1(100)	-	-	-
Total	20	15(75)	3(15)	1(5)	1(5)

Not by chance we observe that 0-1 years age group is the most affected, because many congenital malformations may be diagnosed during intrauterine life through ultrasound and others right after birth. Even if there are lots of prenatal diagnostic methods, lots of pregnant women who come from the rural areas do not come on time to check ups, and the medical facilities in these areas do not poses the necessary medical equipment. Thus, 10 cases of congenital malformations come from the rural areas, 7 cases from Bacau County, and three other cases from other urban areas (table 9).

Table 9. Rural and urban distribution and frequency of the congenital malformation cases

Congenital malformations	No. of cases	Number and frequency (%) of cases		
		Urban areas		Rural areas
		Bacău	Other cities	
Congenital malformations of the heart	9	2(22,22)	2(22,22)	5(55,56)
Hydrocephaly	3	-	1(33,33)	2(66,67)
Microcephaly	1	1(100)	-	-
Congenital malformations of the renal system	3	3(100)	-	-
Congenital malformations of the fingers and toes	2	-	-	2(100)
Congenital Megacolon	1	-	-	1(100)
Multimalformation syndrome	1	1(100)	-	-
Total	20	7(35)	3(15)	10(50)

Conclusions

1. In 2006 we have discovered 27 cases of diagnosed and confirmed genetic maladies and congenital malformations in a total population of 4413 subjects who were brought at The County Pediatric Hospital.

2. The three cases of hemolytic anemia appear at the male sex, who come from the rural area, at the 0-1 years and over 10 years group ages.

3. Out of the four mentally retardation cases 3 are males and 1 female, that come from Bacau city and rural areas (3:1) and from *0-1 years* (2), *3-5 years* (1), *5-7 years* (1) age groups.

4. The 20 cases of congenital malformations represent the majority of cases studied, and their area is very wide: Congenital malformations of the heart (9), Hydrocephaly (3), Microcephaly (1), Congenital malformations of the renal system (3), Congenital malformations of the fingers and toes (2), Congenital Mega colon (1) Multimalformation syndrome (1).

STUDIU PE POPULAŢIILE UMANE DIN JUDEŢUL BACĂU, PRIVIND INCIDENŢA BOLILOR GENETICE ŞI ANOMALIILOR CONGENITALE ÎN ANUL 2006

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Cuvinte cheie: maladii genetice, malformaţii cognenitale, profilaxie

Rezumat

Bolile genetice sunt numeroase. Se cunosc peste 10.000 de boli determinate sau condiţionate genetic. Acestea au o mare diversitate, afectează orice sistem de organe şi ca atare se regăsesc în aproape toate ramurile medicinei.

În această lucrare am studiat frecvenţa maladiilor genetice şi malformaţiilor congenitale, pe parcursul anului 2006, pentru copii din judeţul Bacău. Această iniţiativă vine în sprijinul realizării unui *Program naţional de profilaxie a bolilor genetice.*

Băieţii sunt cei mai afectaţi de malformaţii congenitale, fiind reprezentaţi prin 12 de cazuri comparativ cu fetele care prezintă 8 cazuri. Acest raport se datorează şi naşterii mai multor copii de sex