

## DOWN SYNDROME IN BACĂU COUNTY

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**Abstract:** According to a study on a lot of 27,623 children from Bacău county 23 cases of Down syndrome were found – 7 girls and 16 boys. This syndrome has been found at children between 2 months and 10 years old. These cases are coming from the rural environment – 16 cases and from the urban environment – 7 cases. Among the children with Down syndrome, who had also congenital malformations of the heart, it has been noticed that 3 deceases occurred at boys under the age of 1 year.

### INTRODUCTION

Down syndrome or trisomy 21 syndrome is the most frequent and well – known chromosomal disease. The incidence of trisomy 21 has been estimated at 1/650 – 1/704 newborn babies (according to BRAY et. al.,1998), and the frequency of the products of conception having this trisomy is increased to 1/200 from which  $\frac{3}{4}$  are eliminated as spontaneous miscarriages. The disease is more frequent on male children, the sex ratio is 3 boys to 2 girls (COVIC, 2004). Down syndrome can be clinically diagnosed in the neonatal period or at the newborn children, because of the important abnormalities which, event that they can vary from patient to patient, all realize a phenotypical feature which has led to an improper term of “mongols”. Because of the seriousness of this disease, of the “position” of handicap that the affected persons have and also of social implication of this matter – keeping alive a child with Down syndrome is estimated around 250,000\$ (CEBOTARI et. al., 1998) so a great importance must be granted to the prevention of this syndrome by genetic advice and examination. To realize this is very important to study the cases which are in the analysed area.

### MATERIAL AND METHODS

During 1998 – 2003, it has been made researches on a lot of 27,623 subjects represented by children with ages between a couple of weeks and 16 years old from the County Hospital of Peditry Bacău. In order to discover these cases, it has been realized the clinical examination of the patients who have presented 6 features (COVIC, 2004) from 10 features of Down syndrome (MAXIMILIAN, 1996). Depending of the mother’s age – over 35 years old (HUNTER, 2001) and the presence/absence of such cases in the target family (other children born with this disease) for a certainty diagnosis will be ruled some genetical tests the cariotype of the person (BURKE, 2002). The results of these investigations are shown in the tables 1 – 5 and figure 1.

### RESULTS AND COMMENTS

After the research that has been made in the County Hospital of Peditry Bacău, we found and then there were confirmed and reported 23 cases of Down syndrome (table 1).

Table 1. The frequency of cases of Down syndrome

Year	Number of subjects	Cases of Down syndrome	The frequency of cases (‰)
1998	5183	2	0,385
1999	4878	3	0,615
2000	4819	3	0,622
2001	3919	7	1,786
2002	4413	-	-
2003	4411	8	1,813
1998 – 2003	27623	23	0,832

From these cases, a number of 7 are girls and 16 are boys, confirming in this way the information from medical literature which shows that the cases with Down syndrome are more frequent at boys (table 2).

The most cases are recognized at birth or immediately after birth, but there are a lot of cases recognized later because of the neglect of the child’s parents or due to their ignorance. In what regards the distribution of the cases of Down syndrome according to the group of ages when this disease was confirmed and recognized, we noticed that the group of age with the most cases of Down syndrome was 0-1 year (figure 1). These children, because of their phenotypical feature, are under the suspicion of the existence of this disease.

Table 2 The distribution and the frequency of cases of Down syndrome on sexes

Year	Cases of Down syndrome	The number and the frequency of cases (%)			
		girls		boys	
1998	2	1	50	1	50
1999	3	1	33,33	2	66,67
2000	3	1	33,33	2	66,67
2001	7	3	42,86	4	57,14
2002	-	-	-	-	-
2003	8	1	12,5	7	87,5
1998 – 2003	23	7	30,43	16	69,57

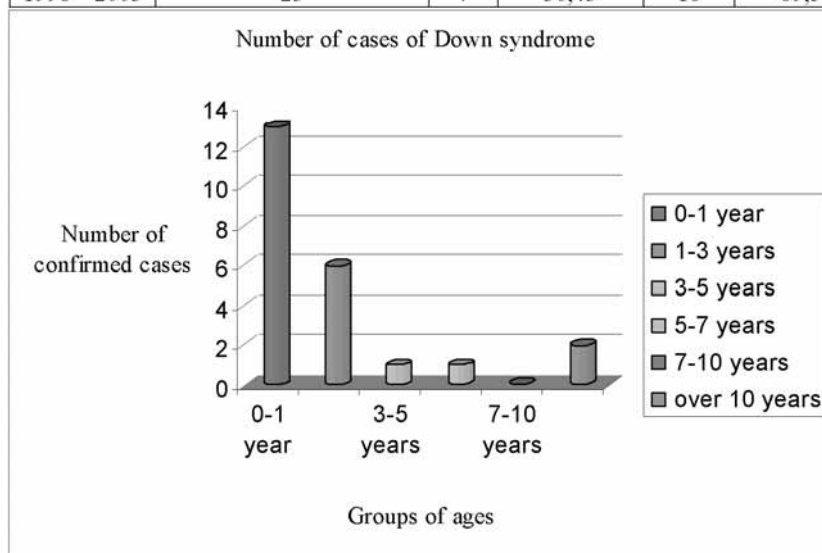


Figure 1 The distribution of Down syndrome according to the ages of children

According to the information from table 3, the 23 cases of Down syndrome found, are classified on groups of ages as following: 0-1 year – 13 cases; 1-3 years – 5 cases; 3-5 years and over 10 years – 2 cases and 5-7 years – one case. It is noticed that regarding group of ages 7-10 years there are attending school and so the incidence of being discovered is increased.

Table 3 The distribution and the frequency of Down syndrome according to the groups of ages

Year	Number of cases	Groups of ages and their frequency (%)											
		0-1 year		1-3 years		3-5 years		5-7 years		7-10 years		over 10 years	
1998	2	1	50	1	50	-	-	-	-	-	-	-	-
1999	3	2	66,67	1	33,33	-	-	-	-	-	-	-	-
2000	3	1	33,33	1	33,33	1	33,34	-	-	-	-	-	-
2001	7	4	57,14	2	28,57	-	-	-	-	-	1	14,29	-
2002	-	-	-	-	-	-	-	-	-	-	-	-	-
2003	8	5	62,5	-	-	1	12,5	1	12,5	-	1	12,5	-
1998 – 2003	23	13	56,52	5	21,74	2	8,7	1	4,34	-	2	8,7	-

The cases of Down syndrome are coming from older parents (especially the mother), have many brothers and sisters, the low level of culture and education of the parents, even if they discover that their child is abnormal (malformations, troubles of reception and speak or mental retardation) and then the parents abandon the children to their grandparents and they didn't put the child in a recovery institution. Also, it is a very serious fact that the child is in impossibility of having medical examination and advice, recovering and mentenance treatment.

According to the table 4, it is noticed that most cases of Down syndrome are coming from the rural environment – 16 cases and from the urban environment are only 7 cases, from which 3 cases are from Bacău city. So, the distribution of the found and confirmed cases with Down syndrome in rural environment is the following: 4 cases in Traian; 2 cases in Nicolae Bălcescu and one case in Mărgineni, Luizi – Călugăra, Parincea, Răcăciuni; Sănduleni; Păncești, Negri, Secuieni, Lipova and Brusturoasa. The distribution in urban environment is the following: 3 cases in Bacău; 3 cases in Buhuși and one case in Comănești.

Table 4 The frequency of Down syndrome in the rural and urban enviroment

Year	Cases of Down syndrome	The number and the frequency of cases (%)					
		Urban environment				Rural environment	
		Bacău		Other cities			
1998	2	-	-	-	-	2	100
1999	3	-	-	-	-	3	100
2000	3	-	-	-	-	3	100
2001	7	1	14,29	4	57,14	2	28,57
2002	-	-	-	-	-	-	-
2003	8	2	25	-	-	6	75
1998 – 2003	23	3	13,04	4	17,4	16	69,56

During 1998 – 2003, there were noticed 3 deceased children with Down syndrome (table 5). The deceases are coming only from the boys under one year, so they died at short time after they were born because of the existence of a congenital malformations of the heart which went to inevitable death. The deceased children are coming from the rural environment – Negri, Luizi – Călugăra and one from the urban environment – Buhuși. These children didn't leave hospital after their birth, being monitorised and investigated till their death occurred.

Table 5 The frequency of deceases in Down syndrome

Year	Cases of Down syndrome	The number and the frequency of cases (%)					
		Urban environment				Rural environment	
		Bacău		Other cities			
1998	2	-	-	-	-	2	100
1999	3	-	-	-	-	3	100
2000	3	-	-	-	-	3	100
2001	7	1	14,29	4	57,14	2	28,57
2002	-	-	-	-	-	-	-
2003	8	2	25	-	-	6	75
1998 – 2003	23	3	13,04	4	17,4	16	69,56

The cases with Down syndrome which have been found, have also a lot of other complications which deteriorate the evolution such as: congenital malformations of the cord – 12 cases; congenital megacolon – one case, malformation of the diaphragm – one case; congenital sprain of the hip – one case and different stages of mental retardation, from which 2 of them very serious ones.

### CONCLUSIONS

During 1998 – 2003 in Bacău county has been found, confirmed and reported a number of 23 cases of Down syndrome among the children between 2 months and 10 years old.

The cases of Down syndrome are distributed on sexes as following: 7 girls and 16 boys who have a severe or a less severe mental retardation.

From all the cases found with Down syndrome, it has been noticed a number of 3 deceases of some children who had congenital malformations of the cord, they were boys of small age coming from both rural and urban environment.

Taking care of the children with Down syndrome raises many problems, both affective and social ones. It is necessary a continuous psychological and adequate support of the parents in order that they can deal with the problems faced in bringing up such a child. So for this reason is very important the realizing of a prenatal diagnosis in order to inform the parents what have they must face and to give them the genetic advice in these circumstances.

### REFERENCES

1. BRAY, I., WRIGHT, D.E., DAVIES, C., HOOK, E.B., 1998. *Prenatal Diag.*, Joint estimation of Down syndrome risk and ascertainment rates: A meta-analzsis of nine published data sets, 18, 9 – 20.
2. BURKE, W., 2002. Genetic testing, *N Engl J Med*, 347, 1867 – 1875.
3. CEBOTARI, L., COTORCEA, V., GROPPA, S., 1998. *Genetica generală și umană. Biopsia corionului în diagnosticul prenatal a patologiilor cromozomale*, F.E.P. Tipografia Centrală, Chișinău, 53 – 55.
4. COVIC, M., 1992. *Genetica umană*, Iași, 86 – 94.
5. COVIC, M., ȘTEFĂNESCU, D., SANDOVICI, I., 2004. *Genetica medicală*, Polirom, Iași, 607.
6. HUNTER, A.G., 2001. *Management of Genetic Syndromes*, Ed. Cassidy SB, Allanson JE, Wiley-Liss, New York, 103 – 129.
7. MAXIMILIAN, C., IOAN, D.M., 1986. *Genetica medicală*, Ed. Medicală, București, 512.
8. MAXIMILIAN, C., POENARU MOȚOC, L., BEMBEA, M., 1996. *Genetica clinică*, Ed. Pan – Publishing House, București, 144.

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