

### Concluzii

1. În anul 2006, au fost descoperite 27 de cazuri de maladii genetice și malformații congenitale depistate și apoi confirmate de specialiști care au fost raportate la Autoritatea Sanitară Bacău dintr-un total de 4413 de subiecți supuși atenției în cadrul Spitalul Județean de Pediatrie Bacău.

2. Cele trei cazuri de anemie hemolitică apar la sexul masculin, ce provin din mediul rural, un caz la grupa de vârstă 0-1 an și 2 cazuri la grupa de vârstă peste 10 ani.

3. Din cele 4 cazuri de retard mental sunt 3 pentru sexul masculin, respectiv 1 pentru sexul feminin, ce provin din municipiul Bacău și din mediul rural (3:1). Au fost depistate la grupa de vârstă 0-1 an (2), 3-5 ani (1) și la grupa 5-7ani (1).

4. *Malformațiile congenitale*, în număr de 20, reprezintă majoritatea cazurilor studiate, iar paleta lor este foarte largă: malformații congenitale de cord (9), hidrocefalie (3), microcefalie (1), malformații de sistem renal (3), malformații ale degetelor (2), meagacolon congenital (1) și sindrom plurimalformativ (1).

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## COMPARATIVE APPROACH IN DOWN SYNDROME EVALUATION - BEGIN BY PHYSICAL THERAPY REHABILITATION

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**Key words:** Down syndrome, B.M.S. system, Portage scale

### Resume

Children who suffer from Down syndrome have difficulties gaining speed when walking, equilibrium, force, eye-hand coordination and gross /fine movement, in comparison with the mentally challenged children.

The measuring instrument called “Basic motor skills test of children who suffer form Down syndrome” (BMS) is a test that measures the motor skills of small children with Down syndrome and is based on the theoretical component “Disturbances in the control of the postural system”. BMS was verified regarding the psychomotricity.

Applying the BMS method as an evaluation technique and as a fundament for the kinetic treatment, allowed to cutback time that was needed for the neuromotor re-education, because the recovery treatment was based especially on obtaining some kinetic fundamental skills, the others objectives being subsumed to this greater one.

### Introduction

Tens of thousands of children suffering from Down syndrome have been studied from the cytogenesis point of view in the last decades. It has become sure that this is the most frequent chromosomal syndrome 1/1700/new born. Then it has become a certainty that there is not one child who has the characteristic malformations of the syndrome and does not have trisomy 21, but in an utterly exceptional way there can be persons who have an extra 21 chromosome but do not present any clinical disorder.

Trisomy 21 is the most common chromosomal anomaly that we encounter in the human pathology. The syndrome is most common in boys, the report being 3 boys to 2 girls.

Recently scientists that take part in the internationally financed Human Genome Project have mapped a succession of approximately 225 genes on the 21 chromosome alone. The research continues in

order to find the genes that are linked to the characteristics of people who have Down syndrome. Once they identify these they may find the biological processes that cause Trisomy 21 thus leading to an intervention or even a cure through gene therapy.

#### *Neurological and motor aspects of children with Down syndrome*

Children who suffer from Down syndrome have difficulties gaining speed when walking, equilibrium, force, eye-hand coordination and gross /fine movement, in comparison with the mentally challenged children.

Cowie (1970) has conducted a study on 97 children with Down syndrome and underlined one of the most characteristic neurological and motor symptoms: hypotonia, visible hypotonia, extreme hypotonia. Alongside these, an important role is played by reduced postural tonus that the child manifests. The reduced postural tonus is associated with insufficient co-contractions, inadequate equilibrium reactions, defected proprioceptive feedback of posture and movement and joint hyper mobility.

The motric development of children with Down syndrome is negatively influenced by the disturbances in the postural control. There are problems in adopting and maintaining the posture and in the movements, that's why the qualitative aspects of movement develop inadequately. Considering all these, it is recommended that we stimulate the development of the movement schemata in children with Down syndrome.

#### *Material and method*

In our research we have applied the recuperation treatment to 3 children who have been diagnosed with Down syndrome.

**Table 1. The structure of the experimental group**

No.	Name	Date of birth	Initial	Final
1.	A.N.	15.10.2006	15.02.2008	15.05.2008
2.	G.L.	21.12.2006	15.02.2008	15.05.2008
3.	M.I.	06.08.2006	15.02.2008	15.05.2008

We have unfolded the experimental action in the kinetotherapeutic office of from "Daniel" Neuromotor Recuperation Centre that pertains to the Association for Supporting the Neuromotor Handicapped Children Bacau.

The duration of the kinetotherapy session for each child was between 30 and 50 minutes.

We have conducted the sessions according to the gravity of the diagnosis form a minimum of 2 sessions/week up to 3 sessions/week.

The measuring instrument called "Basic motor skills test of children who suffer form Down syndrome" (BMS) is a test that measures the motor skills of small children with Down syndrome and is based on the theoretical component "Disturbances in the control of the postural system". BMS was verified regarding the psychometricity.

With this test we can measure the level of postural control of the motor behaviour in the period of basic motor skills development. B.M.S. may be used from the moment that voluntary movement appear until the period when orthostatic behaviour and walking are possible. Generally, the test may be used from three months to three years.

B.M.S. measures the level of postural control for 15 basis motor skills. The skills have been selected from a variety of obvious postural disturbances regarding the postural control, representing the motor problems of the children with Down syndrome in the period of development of basic motor skills. The 15 skills are mentioned in the order that they develop, appear, forming together a growing scale.

In order to validate the theoretical and practical efficiency of the BMS method that we apply mainly on children with Down syndrome, we have utilized for comparison the neurological and motor aspects of the Portage scale for 1 and 2 years (items 46 to 79).

### **Results**

Taking under consideration the fact that the evaluation, for each subject in particular, with the two scales, was very ample, I will present the evolution and the results for the first subject in the experimental group.

For subject *A.N.*, the recuperation treatment led to a favourable evolution of the aspects that we tried to emboss, fact demonstrated by the improvement of the value for each item in the two scales, as presented in Table 2. (for B.M.S.) and Table 3 (for Portage scale).

**Table 2. Evolution of the B.M.S. items in subject A.N.**

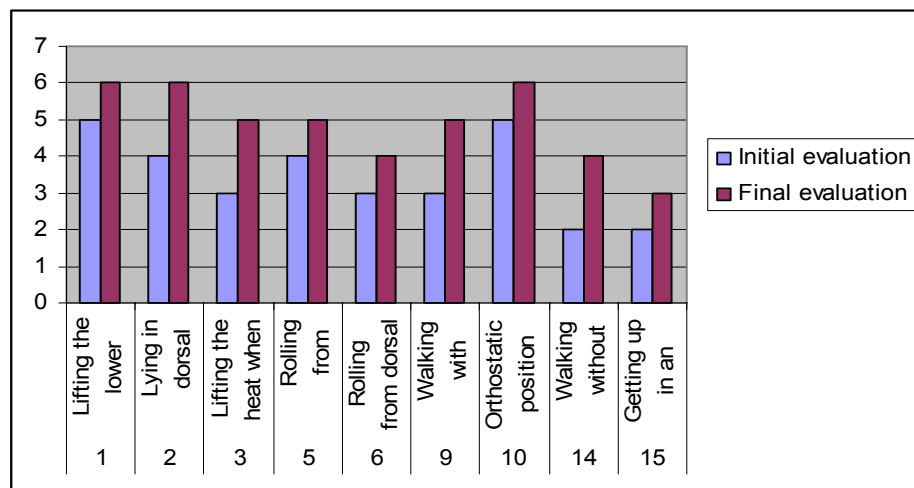
No.	Motor basic skill	Initial evaluation	Final evaluation
1	Lifting the lower members in dorsal decubitus.	5	6
2	Lying in dorsal decubitus.	4	6
3	Lifting the heat when lying in dorsal decubitus.	3	5
4	Standing on the elbows when in ventral decubitus.	4	4
5	Rolling from ventral decubitus to dorsal decubitus.	4	5
6	Rolling from dorsal decubitus to ventral decubitus.	3	4
7	Sitting.	4	4
8	Walking forward on the floor.	4	4
9	Walking with support.	3	5
10	Orthostatic position with support.	5	6
11	Getting up in an orthostatic position with support.	4	4
12	Orthostatic position without support.	4	4
13	Being seated	2	2
14	Walking without support.	2	4
15	Getting up in an orthostatic position without support.	2	3

In B.M.S. system we may notice that with the exception of item 4, 7, 8, 11, 12 and 13, who presented the same value as in the initial evaluation, the other items showed an increase in value with at least one point and more (item 3, 9 and 14). For the items with positive evolution we have presented graphic no. 1.

Into what regards Portage scale for which we have selected 7 representative items, we have found a strong evidence for a very good evolution. This is supported by the fact that at the final evaluation we have obtained the maximum value. (Graphic no. 2).

**Table 3. Evolution of the Portage scale items in subject A.N.**

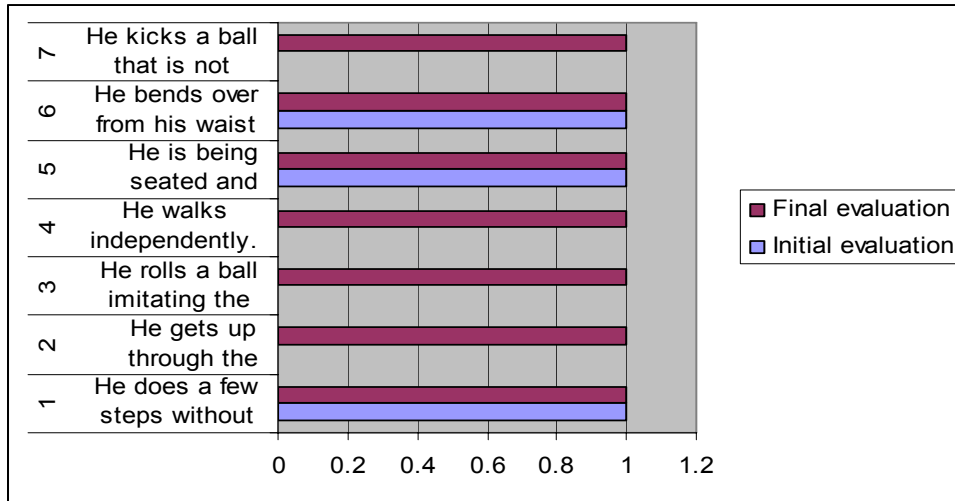
Nr. Crt.	Item	Initial evaluation	Final evaluation
1	He does a few steps without help.	1	1
2	He gets up through the stairs in his knees.	0	1
3	He rolls a ball imitating the adult.	0	1
4	He walks independently.	0	1
5	He is being seated and getting up from this position.	1	1
6	He bends over from his waist in order to lift objects.	1	1
7	He kicks a ball that is not moving.	0	1

**Graphic 1. The item evolution dynamics in B.M.S. method in subject A.N.**

### Conclusions

Considering the theoretical premises that represent the fundament of our work we may argue that our research hypotheses have confirmed:

1. Using motor and somatic-functional examinations through B.M.S. and Portage scale, we have found motor underdevelopment in 3 children with Down syndrome who were a part of the experimental group.



Graphic 2. The item evolution dynamics in Portage scale in subject A.N.

2. Through B.M.S. method (motor development and intervention) we wanted the children with Down syndrome to maintain themselves into the stages of normal development. As a result of our experiment, we found that children with Down syndrome may recover the motor deficit through physical therapy as the main agent of motor education and re-education. Through B.M.S. we have shortened the time that it would normally take to recover the deficit because the treatment was based especially on obtaining a fundamental motor skill, the other objectives being subsumed.

## ABORDAREA COMPARATIVĂ ÎN EVALUAREA SINDROMULUI DOWN - PUNCT DE PLECARE ÎN RECUPERAREA KINETOTERAPEUTICĂ

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**Cuvinte cheie:** sindrom Down, sistemul B.M.S., scara Portage

### Rezumat

Copiii cu sindrom Down dobândesc greu viteza la mers, echilibrul, forța, coordonarea ochi-mână și abilitățile motrice grosiere și fine, comparativ cu copiii cu deficiență mintală.

Instrumentul de măsură denumit „Testul deprinderi motorii de bază ale copiilor cu sindrom Down” (B.M.S.) este un test al deprinderilor motorii special proiectat pentru a testa copiii mici cu sindrom Down și se bazează pe componenta teoretică „Perturbări în sistemul controlului postural”. B.M.S. a fost verificat din punct de vedere psihomotric.

Aplicarea metodei B.M.S. ca metodă de evaluare și de fundamentare a tratamentului kinetic, a permis scurtarea timpului aferent reeducării neuromotorii, deoarece tratamentul recuperator s-a centrat mai ales pe obținerea unor abilități motrice fundamentale, celelalte obiective fiind subsumate acestora.

### Introducere

În ultimul sfert de veac au fost studiați citogenetic mii sau zeci de mii de copii cu sindrom Down. A