"ALEXANDRU IOAN CUZA" UNIVERSITY OF IASI

FACULTY OF BIOLOGY

DOCTORAL SCHOOL OF CHEMISTRY AND LIFE AND EARTH SCIENCES

The Incidence of Certain Genetic Disorders and Congenital Malformations in Human Populations from the Counties of Vaslui and Bacau. Using Physical Therapy Means to Improve a Number of Their Symptoms

SUMMARY OF THE DOCTORAL THESIS

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INTRODUCTION

Genetic disorders and congenital malformations are a current problem of human pathology, due to their frequency, etiopathogenic aspects, and medical-social implications, affecting 5-8% of the newborns and approximately 30% of the individuals during their lifetime.

A few of the ideas expressed by the researchers in this field constituted the reasons why I chose to study the incidence of certain genetic disorders and congenital malformations in the human populations of Bacau and Vaslui, and then to see how much the physical therapy means can contribute to the improvement of some of the disorders' manifestations. By studying the incidence of certain genetic disorders and congenital malformations in human populations of two Moldavian counties, one that is more economically developed (Bacau) and the other one less so (Vaslui), my intention was to observe their genetic health, how it evolved in the last few years, and whether the economical development had any influence on this aspect. This type of disorder - the Down syndrome - was chosen to see how much the physical therapy means can improve the treatment of its sequelae. This disorder was chosen because in the last decades its incidence has grown among the population, drawing the attention of scientists from various fields on its causes, forms of manifestation, and ways of treating the children diagnosed with this syndrome.

PERSONAL CONTRIBUTION
MATERIAL AND METHODS

Research material

To assess the incidence of chromosomal disorders and congenital malformations in humans, I conceived a comparative study, conducted over the course of 8 years on the human populations in two of the Romanian counties (Bacau and Vaslui).

For the experimental part (the physical therapy intervention), the subjects were children with Down syndrome from Bacau county. The experimental study was conducted on a group of 13 subjects (7 males and 6 females), with the clinical diagnosis of Down syndrome, between 6 and 14 years old.

Organization and development of the experimental intervention

The research was conducted between October 2010 and May 2014, in 4 stages, within which tasks, work periods and place were established. The places where the research was conducted were the Didactic and Research Base belonging to the "Vasile Alecsandri" University of Bacau, the Sensory-Motor Integration Laboratory, and the "Down Syndrome Association" of Bacau. The research was conducted over the course of the year 2012-2013, comprising a number of 72 weeks and 360 sessions.

The practical assessment consisted in: somatic measurements, muscle strength measurements, and the tests *"Gross motor function measure"*, *"Clinical Observations of Neuromotor Performance"*, and *"Oseretsky – Guillmann"*, used to assess the children's psycho-motor development during complex activities.

The applicative intervention

The **specific** objectives were to: increase the muscle strength and endurance; educate and rebuild the body image and scheme, the laterality, the spatial-temporal orientation and organization; increase the stability, balance, control, and coordination.

The physical therapy programs comprised:

- Various ways of triggering motor acts and actions;
- Maintaining the postures and positions and performing active movements during it;
- Continuing the already known movements;
- Using alternating global and segmental movements, proprioceptive neuromuscular facilitation, therapeutic physical exercises during applicative tracks;
- Improving the stability, the balance, and the perceptive-motor coordination throughout the program, going from static to dynamic balance;
- Being more aware of the body scheme, laterality, and temporal-spatial orientation;
- Improving the skills during the daily life activities.

RESULTS OF THE RESEARCH

The statistical analysis of the data collected from the Public Health Directions showed that the incidence of the chromosomal disorders recorded in the human populations of the Bacau and Vaslui counties over the course of the period that was analyzed had average values of 0.014% in Bacau county and higher, of 0.025% in Vaslui county, presenting important variations from one year to another, as follows: between 0.0004% (2009) and 0.0054% (2013) in Bacău county, and between 0.0009% (2008) and 0.012% (2013) in Vaslui county.

The chromosomal disorders represented 9% of all the anomaly cases (chromosomal and congenital) that were identified for the studied period in Bacau County and 13% of them in the county of Vaslui. In both counties there was a higher frequency of chromosomal disorders in 2013, the spotted cases representing, out of the total cases identified throughout the entire period, 38.14% in Bacau County, and 47.17% in Vaslui. The incidence of congenital malformations was also higher in Vaslui (0.024%) than in Bacau (0.020%),

oscillating between 0.008 - 0.037% in Vaslui County, and between 0.008 - 0.047% in Bacău.

In regards to the sex distribution of the chromosomal disorders and congenital malformations, in the human population of Bacau county, these disorders affect about the same way both sexes (49.62% female cases and 50.38% male cases), while in Vaslui county, the number of male cases are a bit higher than the female ones (46.08% female cases and 53.92% male cases).

In regards to the age distribution of the investigated disorders, in Bacau County they were diagnosed in the age group of 0-1 year old, while in Vaslui County, most of these disorders were spotted in the age group of 1-14 years old. It is surprising that in both counties an important number of malformations were diagnosed in the age group of 15-64 years old.

Following the physical therapy intervention over the course of approximately 1 year and a half on the Down syndrome subjects, I noticed that the psycho-motor deficiencies recorded initially have obviously diminished. Somatometry shows a growth and development in relation to the children's age due to natural development over the course of time, but also due to the physical activity performed almost daily. This is also emphasized by the somatic measurements and psycho-motor tests that were applied.

The analysis of the *"Gross motor function measure"* (GMFM) test results offered us concrete data with a reduced margin of error, because the assessment consisted of simple, known actions that were implemented in children in the first stages of the basic motor skills education. The initial testing emphasized the level of functional development, during static and dynamic activities, the values being between 47.05% and 59.52% (out of 100%), while in the final testing the maximum values were between 99.04 % and 100%.

After analyzing the results recorded by the entire group of subjects for the test *"Clinical Observations of Neuromotor Performance"*, I observed that by using the physical therapy intervention, the *"sensory perception"* and *"postural reactions"* have improved by 4 points out of a total of 8, the *"bilateral motor abilities"*, by 5 points out of 6, and the criteria *"somatopraxis"* and *"other clinical observations"* by 2 points out of 4. The great majority of the subjects have improved their neuro-motor results by over 80%, the smallest progress being recorded by subject 9 (an improvement of 54.8%), while the biggest progress, by subject 5 (96.77%). The results for the *"Oseretsky"* test revealed that following the initial and final tests of the gross motor skills there was a difference in the arithmetical mean of 74.77 points, representing the progress recorded for the 4 psycho-motor components - *Speed and agility, Balance, Bilateral coordination*, and *Strength*. In regards to the *fine motor skills*, the initial arithmetical mean was of 36.46 points, and the final one, of 107.07 points, which shows the development of the following skills: *response speed, visual-motor control, speed, and dexterity*. After the physical therapy intervention, the *Perceptive-motor coordination* has improved by 16.61 points.

CONCLUSIONS

- I believe that the higher incidence of chromosomal disorders and congenital malformations in the human population of Vaslui county does not depend so much on the eating habits (which are probably healthier in this county, its rural population being higher), nor on the pollution level of the area (more reduced than the one in Bacau - a more industrialized county), but more likely on the county's level of development, the financial state of its population, civilization degree, its probably more precarious medical assistance, the level of education and instruction of the future mothers, etc.
- The issue of diminishing the incidence of chromosomal disorders and congenital malformations remains a very current one for the local and central health commissions in charge of ensuring the genetic health of the human populations. Through this scientific study I hope to have contributed to the awareness of certain realities in two of the Romanian counties. At the same time, through the physical therapy program, applied individually and on groups of patients with Down syndrome, there was an improvement in several psycho-motor skills of the subjects, such as muscle strength, laterality, body scheme, stability, balance and coordination, this contributing to an obvious improvement of the quality of their lives.

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