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IMPROVING THE QUALITY OF LIFE FOR CHILDREN DIAGNOSED WITH OSTEOGENESIS IMPERFECTA THROUGH EXERCISES IN THE AQUATIC ENVIRONMENT

DOCTORAL THESIS
ABSTRACT

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PART I, **THEORETICAL FRAMEWORK OF THE RESEARCH**, contains 5 chapters:

Chapter 1: Introduction to the research topic; **Chapter 2**: Considerations regarding the reflection of research topic in the national and international literature; **Chapter 3**: General aspects relating to osteogenesis imperfecta; **Chapter 4**: The aquatic environment and exercises in the aquatic environment; **Chapter 5**: Adapted sport and its implications in the physical activity of children with disabilities.

Motivation for choosing the theme

Consequent to the fact that the specialized current research is mainly oriented towards the medical field, including the surgical one, and the specific medication for this disease, we consider it appropriate to conduct a study for the implementation of a physical exercise program in the aquatic environment, in order to increase the quality of life for people with osteogenesis imperfecta (OI) or

Lobstein's disease. As a major research area, we are referring to the physical education and sports field.

Purpose of the work

By achieving this work, we aim to implement an aquatic exercise program adapted to the needs and abilities of the people concerned, also including the use of aqua bike.

Osteogenesis imperfecta or Lobstein's disease, given its particularities and associated disabilities, is a challenge for both the medical field and the physical education field towards improving the quality of life for people diagnosed with this condition.

Through this research, we aim to make known a case study conducted on two Romanian subjects suffering from Lobstein's disease, in order to demonstrate the evolution and the functional and mental motor progress of the investigated subjects after completing a program in the water environment. We believe that the conducted research can contribute to developing methodical intervention possibilities of improving the movement ability in people diagnosed with OI, from the perspective of sports science and physical education field.

Osteogenesis imperfecta or Lobstein's disease is a rare, inherited disease of the connective tissue, characterized by weak bones. It is a disease caused by an anomaly in the production of 1A1 collagen and 1A2 collagen, the main protein in the bone. Type 1 collagen fibers can support heavy loads while allowing a certain degree of mechanical deformation. OI equally affects the bone quality and its mass, being represented by the constant emergence of fractures. People diagnosed with OI encounter medication-related problems all their life long, but its treatment is being continuously enhanced.

This disease occurs in 1/15000 - 1/20000 people, regardless of populations and gender. The estimate does not include the mild forms of the disease, which may go unnoticed. Worldwide, there are affected around 500,000 people, namely only 0.008% of the global population.

In Romania, the specialized literature does not provide studies relating to the assessment, in physical terms, of an adapted physical program or the systematization of means to improve the quality of life for people diagnosed with OI. As regards the specialized literature at a global level, we can state that the studies focused on improving the quality of life for people diagnosed with OI have been mainly directed towards the medical field and very few of them towards the physical education field.

The article *Exercises in osteogenesis imperfecta*, achieved on 9 September 2013, in Brazil, is the most extensive research at the international level, aimed at collecting and systematizing all studies on the physical activity of people with osteogenesis imperfecta.

Water is the most recommended environment to carry out physical rehabilitation or recovery programs, when speaking about people diagnosed with OI. It is an ideal environment due to the characteristics of the water, taking into account the particularities involved by Lobstein's disease, the risk of fractures being thus very low if the program is performed under the guidance of a therapist.

Because the aquatic environment is not yet maximally exploited, it presents a high potential for recovery and rehabilitation. Specialized studies confirm and support the aquatic environment due to the fact that it provides high security for the programs carried out and technical adaptability as well.

Hydrokinetotherapy (HKT) is not only the transposition of medical gymnastics or kinetotherapy programs from the dry land to the water environment, but it involves the exertion of joints, which first of all improves flexibility and secondarily aims at a muscular effect.

Therapeutic swimming is part of HKT and ensures the long-term functional maintenance of the body, having multiple therapeutic indications in almost all areas of pathology, as well as prophylactic indications, serving to treat various diseases of the musculoskeletal, cardiovascular and respiratory systems.

Besides the techniques applied in HKT and therapeutic swimming, we recommend people with OI to use, as adjuvant methods in the recovery process, running into deep water or water running, aqua cycling and water games.

PART II, **PRELIMINARY STUDY ON THE IMPROVEMENT OF MOVEMENT ABILITY IN THE CASE OF A SUBJECT DIAGNOSED WITH OSTEOGENESIS IMPERFECTA**, includes 3 chapters:

Chapter 6: Organizing the preliminary study; **Chapter 7**: Exploration of a presumptive aquatic program proposed to improve the quality of life for children with osteogenesis imperfecta; **Chapter 8**: Conducting the preliminary study.

Purpose of the study

The preliminary study aims to track the effects induced by the application of an aquatic program designed to improve the quality of life in the case of a subject diagnosed with OI.

Objectives of the study

- ✓ Optimizing the movement ability of a child diagnosed with osteogenesis imperfecta;
- ✓ Designing an aquatic program able to improve the quality of life of a subject diagnosed with osteogenesis imperfecta;
- ✓ Developing interest in the practice of swimming and aquatic activities.

The preliminary study was based on the application of an aquatic program designed to improve the quality of life in the case of a subject diagnosed with OI. The program was constructed according to the methodical principles applied to adapted activities: principle of individualization, principle of accessibility, principle of conscious and active participation, principle of solid learning and durability of the achieved results, principle of gradual effort or progressive adaptation to the programmed type of demand, principle of motivation.

The aquatic program used in the preliminary study was conducted in two stages. Stage I covered the weeks 1 to 5, during which it was tracked the awareness and habituation with the water environment. Stage II included the weeks 5 to 30, where the focus was on the improvement of exercise and movement capacity and the development of personal motivation.

In achieving the program, it was observed the basic structure of any other physical program: preparing the body for effort, performing the exercise in the fundamental part and cooling down after exercise in the final part.

Stage II of the program was structured in three essential parts: technical training with an emphasis on mobility, endurance training and strength training. Experts recommend that the three directions (range of motion, including joint mobility, endurance and strength) developed within the mentioned programs should represent the basis of any program for people diagnosed with OI.

The assessment techniques used in the preliminary study were the following: anthropometric assessment of the range of motion, muscular testing, assessment of movement ability through the specific sheet, assessment of the quality of life using the QOLS questionnaire (The Quality of Life Scale), assessment of psychological well-being using the RYFF questionnaire (The Ryff Scales of Psychological Well-Being) and osteodensitometry.

Discussion on the preliminary study results

After performing the joint and muscle assessments at the Shriners Hospital, in Canada, we can assert that the subject has undergone an increase in muscle strength at the level of lower limbs. It is worth mentioning that during the entire period when the aquatic program was applied, the subject did not suffer any other fracture of the lower limbs.

Bone density of the subject has increased by 12.7%.

After completing the sheet for the movement ability of the subject, we have noted major improvements in all actions performed by him in the water environment. At the end of the aquatic program, he had managed to learn all the basic technical elements – floating, sliding and, most important, water breathing. Also, he had learned the technique of crawl, backstroke and butterfly swimming, as well as the breaststroke arm movement, all of them representing a very strong means to fight for improving the quality of life. He had also learned the underwater swimming technique and the turnover technique.

At the level of lower and upper limbs, the subject has undergone an increase in muscle strength, which is proven by his ability to perform, both assisted and independently, a greater number of squats, lifts on tiptoes, as well as push-ups with support on knees or support on the edge of the pool.

As for the torso, the subject has undergone muscular improvement, which is revealed by his ability to hold the isometric position from support on the knee and forearm.

In terms of endurance, after completing the 20 weeks of the program, the subject was able to swim 500m continuously using the crawl stroke and he also managed the personal performance to run into deep water for 10 minutes.

Psychological questionnaires have shown considerable improvements at all levels.

The preliminary study results allow us to develop in the final research the preparation program in the water environment by complementing it with a new means designed to improve exercise and movement capacity, namely the aqua bike.

PART III, **RESEARCH ON IMPROVING THE QUALITY OF LIFE FOR CHILDREN DIAGNOSED WITH OSTEOGENESIS IMPERFECTA THROUGH EXERCISES IN THE AQUATIC ENVIRONMENT**, contains 4 chapters:

Chapter 9: General framework for organizing the research; **Chapter 10:** Conducting the research; **Chapter 11:** Results and their interpretation; **Chapter 12:** Designing and building the aqua bike.

Purpose of the research

Through this final part of the research, we aim to improve the quality of life of two subjects diagnosed with osteogenesis imperfecta by applying an aquatic program, for one of the subjects also using the aqua bike, as a complementary means.

Objectives of the research

- ✓ Developing exercise and movement capacity in subjects diagnosed with osteogenesis imperfecta;
- ✓ Improving the psychological factor and motivating the subjects to improve their quality of life.

The research hypotheses are:

- ✓ The aquatic program leads to an increase in bone density of the participating subjects;
- ✓ The aquatic program that also uses the aqua bike leads to increased exercise capacity;
- ✓ An efficient way of practical and methodical intervention is the use of aqua bike.

The research was conducted on two subjects of almost similar ages (11 and 12 years), classified with type IV OI, and it took place from May 2014 to March 2015; we mention that one of the two subjects (subject II) has a relative classification, between type III and type IV, according to the diagnosis established by the "Orthopadisches Spital Speising", a hospital from Vienna, Austria.

The assessment techniques used were the following: anthropometric assessment performed with the Tanita SC-240-MA Body Composition Monitor, assessment of the quality of life using the QOLS questionnaire (The Quality of Life Scale), assessment of psychological well-being using the RYFF questionnaire (The Ryff Scales of Psychological Well-Being), assessment of movement ability, osteodensitometry and spirometry.

Implementation of the aquatic program model

In the final research, it was used the aquatic program applied during the preliminary study and it was thus developed stage III of the aquatic program.

The aquatic program from the preliminary study was applied to subject II, in order to confirm or not its effects on the quality of life of children diagnosed with osteogenesis imperfecta.

Given the constitutional typologies of the two subjects, in the case of subject I, the emphasis was placed on increasing muscle strength and mass, while in the case of subject II, the focus was on decreasing the percentage of body fat and improving muscle tone.

For subject I, it was applied stage III of the aquatic program developed in order to continue the preliminary study program. This one was designed taking into account the methodical principles aforementioned.

Because the application of the aquatic program conducted in three stages considered the progressive exercise parameters, it was highly safe for the subjects.

Stage III included the ***weeks 30 to 60*** and had the following objectives:

- To increase bone density;
- To improve respiratory functions;
- To increase muscle mass and decrease body fat levels;
- To increase motivation by obtaining physical progress.

Stage III of the aquatic program followed the structure of stage II, being divided into three parts: technical training, endurance training and strength training.

Results obtained

After completing the aquatic program, the subjects have shown increased bone density and improved movement ability.

In psychological terms, after assessing the two psychological questionnaires, there have been recorded increases at all levels.

In the case of subject I, who followed all three stages of the program, the spirometric examination has indicated increased breathing capacity.

Designing and building the aqua bike

Aqua bike represents our contribution to the fight against this ruthless disease. As in the case of running into deep water, it is used as a means to develop exercise and movement capacity in people with Lobstein's disease.

Aqua bike is a metallic construction made of alimentary stainless steel, in order to resist the acidic conditions existing in the working environment. Both its subsets and the moving bodies are special, being protected so as to exclude contact with the working environment.

The innovative particularity of aqua bike is represented by the possibility to tilt it laterally, which engages the upper body of the user during exercise. The construction is equipped with limitation to tilting, which eliminates the danger of sideways overturning.

General conclusions

- ✓ Osteogenesis imperfecta or Lobstein's disease, given its particularities and associated disabilities, is a challenge for both the medical field and the physical education field towards improving the quality of life for people diagnosed with this condition.
- ✓ During the application of the aquatic program, subjects did not suffer any fracture, but instead they have undergone a significant increase in bone density.
    - ***In connection with the above statement, we consider that hypothesis no. 1 of the research,*** *"the aquatic program leads to an increase in bone density of the participating subjects",* ***has been validated.***
- ✓ After completing stage III of the aquatic program, it has been achieved an improvement of respiratory functions in the case of subject I.
    - ***In connection with the results of spirometric examination for subject I, we consider that hypothesis no. 2 of the research,*** *"the aquatic program that also uses the aqua bike leads to increased exercise capacity",* ***has been validated.***
- ✓ The aquatic program has brought improvements in the muscle mass and fat mass of the participating subjects.
- ✓ Aqua bike has become an efficient means of the applied program, being used in its last part, obviously without the emergence of injuries. Following the assessments performed at the muscle and respiratory levels, there were recorded positive results, which have led to an increase in the quality of life of the participating subjects.
    - ***In connection with the above statements, we consider that hypothesis no. 3 of the research,*** *"an efficient way of practical and methodical intervention is the use of aqua bike",* ***has been validated.***
- ✓ The aquatic program represented a starting point for the international classification of one of the subjects. He was framed in the category S8SB7SM8, already participating in international competitions in Canada.

Contributions and practical applications of the research

- ✓ Achievement of the aquatic program aimed to improve the quality of life for children diagnosed with osteogenesis imperfecta;
- ✓ Design of a personal sheet for the movement abilities of children diagnosed with osteogenesis imperfecta, which can become an essential assessment method within any aquatic program aimed to improve the quality of life;
- ✓ Development of a compendium of exercises with minimal risks for people diagnosed with OI, which can be the basis of any aquatic program designed for osteogenesis imperfecta;
- ✓ Building the aqua bike and its implementation in the aquatic program for children diagnosed with osteogenesis imperfecta;
- ✓ After completion of the aquatic program, subject I was designated as a "Hopeful Athlete in Para-Swimming" and was elected to bear the flame of the Quebec Games, in Canada, at the opening of the *Défi Sportif* competition;
- ✓ An official letter was received from Claude Picard, Head Coach of the Para-Performance Swimming Club in Canada, through which he was appreciating the very high level of subject I at the moment of his integration into the Paralympic team of the mentioned club. Picard was also appreciating the mastery of the subject in the technique of all four swimming styles, as well as his muscular level. It was highlighted that, from the psychological point of view, the subject had a high degree of autonomy, was very motivated about his future sports activity and had maximal self-confidence. Not least, Claude Picard specified that the subject was reclassified from the category S9 SB8 SM9 to S8SB7SM8, which would allow him to progress and participate in the Paralympic Games;
- ✓ Subject I participated in the first Paralympic regional competition in Canada, with excellent results represented by 6 medals (a gold medal in 400m freestyle, 3 silver medals and 2 bronze medals).