

STUDIU DE CAZ PRIVIND EFICIENȚA UNUI PROGRAM KINETIC PERSONALIZAT ÎN SINDROMUL RETT

CASE STUDY REGARDING THE EFFICIENCY OF A PERSONALIZED PHYSICAL THERAPY PROGRAM IN RETT SYNDROME

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Abstract

Introduction. Rett's syndrome is a genetically caused neurological disorder which affects mainly girls. It is characterized by normal initial development followed by the slowing down of development, loss of ability to use the hands, distinctive movements, slowing down of head and brain growth, gait disorders, seizures and intellectual problems.

Purpose. The aim of this paper work is to study the role of physical therapy combined with Bowman therapy in supporting the neuro-motor development of the patient suffering of Rett's syndrome and the improvement of their function, by accomplishing a case study.

Material and method. The study was performed on a female subject aged 4 years and 8 months old, diagnosed with Rett's syndrome. The assessment means used were: Gross Motor Function Classification System, scoliometer, anamnesis, interview with parents. As therapeutic means, the followings were used: physical therapy, Bowman therapy, oxygen-therapy, dondolino.

Results. Weight and height within normal limits, severe neuro-motor retardation, high discrepancy between biological age (2 months) and the chronological one (4.2 years), 5° scoliosis. After 8 months of physical therapy and Bowman therapy, the biological age ameliorated with 2 months, the gastro-intestinal and sleeping disorders were still present but ameliorated after treatment.

Conclusions. A personalized physical therapy programme, combined with complementary therapies, can have beneficial effects upon the neuro-motor and functional development of the child suffering of Rett's Syndrome.

Key words: Rett's syndrome, personalized programme, physical therapy, Bowman therapy, oxygen therapy.

Rezumat

Introducere. Sindromul Rett este o afecțiune neurologică cauzată genetic, ce afectează în special fetele. Este caracterizat printr-o dezvoltare inițială normală, urmată de o încetinire a dezvoltării, pierderea abilității de utilizare a mâinilor, mișcări distinctive, încetinirea creșterii capului și a creierului, probleme de mers, crize și probleme intelectuale.

Scop. Lucrarea își propune să studieze rolul kinetoterapiei combinate cu terapia Bowman în susținerea dezvoltării neuromotorii a pacientei cu Sindrom Rett și îmbunătățirea funcției acestora, prin realizarea unui studiu de caz.

Material și metodă. Studiul s-a realizat pe un subiect de gen feminin, în vârstă de 4 ani și 8 luni, diagnosticată cu sindrom Rett. Mijloacele de evaluare folosite: Gross Motor Function Clasification System, scoliometrul, anamneza, interviul cu părinții. Ca mijloace terapeutice s-a utilizat kinetoterapie, terapia Bowman, oxigenoterapie, dondolino.

Rezultate. Greutate și înălțime în limite normale, retard neuromotor sever, discrepanță mare între vârstă biologică (2 luni) și cea cronologică (4,2 ani), scolioză de 5°. După 8 luni de kinetoterapie și terapie

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Bowman, vârsta biologică s-a ameliorat cu 2 luni, probleme de somn și gastrointestinale prezente dar ameliorate după tratament.

Concluzii. un program de kinetoterapie personalizat, combinat cu terapii complementare, poate avea efecte benefice asupra dezvoltării neuromotorii și funcționale a copilului cu Sindrom Rett.

Cuvinte cheie: sindrom Rett, program personalizat, kinetoterapie, terapie Bowman, oxigenoterapie

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INTRODUCTION

Rett's syndrome is characterized by a normal initial development followed by a slowing down of development, loss of ability to use the hands, distinctive movements, slowing down of head and brain growth, gait disturbances, seizures and intellectual problems. Apraxia – the lack of ability to perform motor functions – is probably the most severe consequence of Rett's syndrome, which interferes with every move of the body, including speaking and watching. (Zaa-A Han and co., 2012).

As symptoms, we can also mention: walking on tip toes, sleeping disorders, delayed growth, seizures, cognitive disabilities, and respiratory disorders while awake, such as hyperventilation, apnea and air swallowing (D.C. Tarquinio and co., 2010).

Other symptoms, which can vary from slight manifestations to severe ones can be: respiratory problems – the problems seem to become worse under stress conditions; usually, breathing is normal during sleeping and abnormal while awake; changes in development, excessive saliva, fidgety hands and feet – frequently this is the first sign, intellectual disabilities and learning difficulties; scoliosis, seizures, loss of social interaction, severe continuous constipation and gastro-esophageal reflux, poor circulation, which can lead to cold and blue hands and feet, severe problems in language development. (Naidu, S., 1997)

M. Shevell, MD and collaborators, in their study “Assessment of the child with global development delay” had as purpose the achievement of certain recommendations based on evidence regarding the assessment of the children with non-progressive delays in their global development. Due to the increased incidence of visual and hearing disabilities, children with delay in their global development should also benefit of visual and audiometric assessments when they are diagnosed (M. Shevell, MD, S. Ashwal, MD and co., 2003). The understanding of the cause of this disorder is required for the development of new therapies and to control specific symptoms, as well as to provide better diagnosing methods.

Although associated with a devastating loss of function at an early age, until the age of 5, its evolution becomes relatively static after this age, fact which determines the position of Rett's syndrome far away from most neuro-degenerative genetic disorders of childhood (Lane J.B and co., 2011).

The treatment for this disorder is symptomatic – focusing on managing the symptoms – and supportive, which requires a multidisciplinary approach. Medicines can be administered for the respiratory irregularities and motor difficulties and anti-convulsive medicines can be provided to control seizure. Occupational therapy can help children develop their abilities necessary to perform self-directed activities (such as dressing up, feeding, arts and crafts), while physical therapy and hydrotherapy can help their mobility (Naidu, S., 1997). The symptomatic treatment could include: assistance at feeding and other ADLs (daily activities), methods for treating constipation and gastro-esophageal reflux, physical therapy to help prevent hands problems, exercises to control weight for those with scoliosis. Administration of food supplements can help those with slow growth.

AIM

The purpose of this paper work is to study the role of physical therapy combined with Bowman therapy in supporting the neuro-motor development of the patient suffering of Rett's Syndrome and improvement of function by the accomplishment of this study.

HYPOTHESIS

A personalized physical therapy programme, combined with complementary therapies, has positive effects in the neuro-motor and functional development of the child suffering of Rett's Syndrome.

MATERIAL AND METHOD

Subjects. The subject used in this case was C.C.M., a 4 years and 8 month old little girl, born on the 10th of August, 2009. She resulted from a normal pregnancy, birth through caesarian operation because it was depicted a fetal hypotrophy, placental insufficiency and its aging. Her weight at birth was 2,200 grams, her length 44 centimeters and her Apgar score was 8/9. Initially, at the age of 8 months, she was diagnosed with cerebral palsy, lax paraparesis, having as secondary disorders a neuro-motor retardation, then she was diagnosed with lax tetraparesis and then, only at the age of 2, she was suspected of suffering of Rett's syndrome, this diagnosis being confirmed in Hungary, in Debrecen, in January 2012, when the patient was 2 years and 5 month old.

The neuro-motor development was at first slowed down, she never sat down and she never rolled properly. A delay in her neuro-motor development was interpreted, for which she was subjected to physical therapy and after that, a slight amelioration was noticed (in VD she was making efforts to hold her head, she was supporting herself on her arms and hands).

During that period she was holding her nursing bottle with both hands, she was grabbing toys, she was transferring them. Starting with the age of one and a half, the patient lost prehension and after the age of 2, apprehension was lost, but she played obsessively, pulled her hands, agitated them on the medial line, took them to her mouth, liked them, sometimes bit them and contact was no longer possible with her. She did not execute any order, she was completely detached.

In May, 2011, she started having seizures, initially from sleep, which manifested themselves through stare, involuntary movement of upper limbs and later, some seizures during the day too. The seizure in September manifested itself with clonuses at upper limbs, stare and duration under one minute.

Assessment tools

In the studied specialty paper works, the most frequently used assessment means are based on observation, anamnesis, questionnaires with questions for families, or various medical investigations, such as imaging or laboratory ones.

a) GMFCS (Gross Motor Function Classification System): see Annex 2. GMFCS in infantile neurological disorders is based on the assessment of independent movements, the emphasis being laid on the assessment of sitting, transfer and mobility abilities. In defining this 5 degree classification system, it was considered of utmost importance the fact that the differences between these degrees should be significant in habitual life. It is important that in assessing the child's motor functions, the emphasis should be laid on her gross motor abilities and not on her disabilities.

b) Postural assessment: for this, the inspection method was used and the **scoliometer**. The subject was placed in sitting position, ventral decubitus, respectively dorsal decubitus, while the assessor, depending on what the purpose was in the respective moment, in the front, behind, homolateral or heterolateral to the subject. It was observed the correctness of body alignment, the presence or absence of posture deficits and if they are of structural or functional type.

c) Anamnesis: the family were asked various questions related to the patient's medical history, to the development and treatments that she has benefited of until present, respectively to their results and effects. Through this method, there were also identified the patient's disorders related to sleeping and gastrointestinal system. The conclusion of anamnesis will be presented when describing the patient in the discussion section.

Medication. In the present, the patient is being administered only Depankine syrup, being dependent on this medicine, which keeps her seizures under control and prevents their reoccurrence. Moreover, if necessary, she is also administered medication for constipation. While she was hospitalized for epileptic seizures, she followed a treatment with PEV, Manitol, HHC, Depakine, Levetiracetam, Tanakan, Rivotril.

Personalized physical therapy programme. After the initial assessments and taking into consideration the parents' options, the following general objectives were established: stimulation of neuro-motor development; achievement of head, neck and upper body control; prevention of generalized articular ankylosis; stimulation of circulatory and respiratory systems; amelioration of constipation; influencing the sleeping patterns; maintaining the sitting position; stimulation of preparation for jump reflex.

The physical therapy sessions were carried on twice per week, lasting about 40-60 minutes each. The average of exercise dosing was 3 series per exercise of 10 executions for each of them. Present physical therapy was begun in November 2013 and it still continues. As means, the following were used: neuro-motor facilitation, stretching, the big fitness ball, the mattress, the physical therapist's grips and counter-grips (Căciulan Elena, Stanca Daniela, 2011; Ciobanu Doriană, 2011). After this exercise programme, a few relaxation and loosening techniques will be executed: shaking, passive mobilizations and tractions, passive stretching.

Ex. 1: P.I.: DD with LL flexed, feet on the mattress. The physical therapist fastens the knees with her hands and slightly „pulls” the quadriceps muscles to determine patient to lift her buttocks from the ground. The purpose is to achieve the contraction of gluteal muscles, even though the patient only manages to initiate the move, without fully executing it.

Figure 1. Exercise 1



Ex. 2: P.I.: DL, UL slightly flexed, the UL from above is situated along the trunk.

The physical therapist fastens the ground UL with one hand and with the other one she executes the hold on the shoulder above, facilitating the lateral head lift. The exercise is repeated on both sides.

Figure 2. Head lift to the left



Ex. 3: P.I.: The patient is placed in lateral support on her forearm, the other hand being on the mattress. The physical therapist supports the patient from behind and places one hand on the lateral mattress side of the trunk. The purpose of this exercise is loading on the forearm and determining the patient to support herself on the free hand when she loses her balance as a result of trunk muscles instability. Another aim is for her to maintain her head on the medial line.

Figure 3. Exercise 3



Ex. 4: P.I.: DD on the mattress. The physical therapist is in front of the patient, holds her hands and partially lifts her until the head is no longer supported on the mattress/pillow and she maintains the position until the patient raises her head, bringing her chin towards her chest. After that, the physical therapist lifts her to sitting position and maintains it as much as possible.

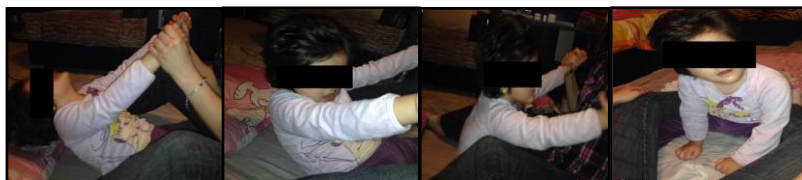


Figure 4. Getting from dorsal decubitus into sitting position

Ex.5: VD on the big fitness ball, hands behind the back above the pelvis, head outside the support area. The physical therapist, with elbows on the ball, supports the patient's shoulders, so as she

would raise her head on the medial line and be in normal alignment from anatomic point of view. The patient executes head extension and flexion.

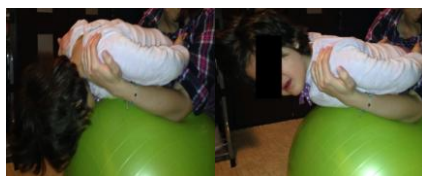


Figure 5. Head extension and flexion

Bowman Therapy. It is a unique formula of neuro-muscular rebalancing.

Bowman therapy is a new concept in body manipulation, it does not derive and neither is similar to any other way of physical approach: bone movements are realigned, muscular tensions are released and the normal lymphatic circuit is reestablished, the energetic meridians are immediately reestablished, psychic balance is normalized. The duration of a session is between 40 and 60 minutes. A Bowman therapy session consists in a number of delicate moves performed with the fingers upon muscles and articular tissues. Thus, messages are transmitted inside the body, messages of cellular memory reactivation, reactivation of an ideal condition of relaxation and balance (www.bowtech.ro, 23.06.2014).

Dondolino device. Dondolino is a standing frame which was created to help children aged 1 to 10 to achieve and maintain an independent orthostatic position; to maintain a vertical position with loading on both lower limb, a an alternative for those who cannot achieve this independently. It is important to remember the fact that the orthostatic position is the ideal antigravitation posture for people. It represents the support on the two lower limbs, allowing thus the upper limbs to be free, without having to act as support and protection. Dondolino is an aid which allows these pieces of information to be integrated and facilitates this process (Use and maintenance handbook, 2014).

This device was purchased for the patient when she was 3 years old. At the beginning she resisted for a few minutes but as she started to grow she could not be verticalized anymore as she would yell or cry as soon as she was placed in orthostatic position or when they tried to verticalize her. That was why they gave up on the device.

Oxygen therapy. The requirement of therapies is about 15-20 sessions per treatment series (depending on the patient's response to therapy and her cooperation). It is desired that such a cycle should be carried on during a minimum of 15 sessions, but no more than 20 sessions per series. The scientific studies performed in this respect show that a cycle of about 40 hours of hyperbaric oxygenation creates the conditions to make efficient the conditioning therapies. The hyperbaric oxygenation therapy is performed on a rate of one session per day, seven days per week. The patient was subjected to the first session of hyperbaric oxygenation during the 24th of October, 2011 and the 7th of November, 2011, representing 15 sessions performed according to scheme TS 240/90 (cf. ECHM), her mother, C.A., being her assistant. The therapy was carried on without complications, being noticeable an improvement of the patient's attention, fact which makes us hope in a progressive rehabilitation together with the associated therapies.

RESULTS

Anthropometric measurements (height and weight)

As comparative element, it was used the standard growth in height graphic of the child since from birth till the age of 8. We have to mention that these are average, orientation values. There are no perfect delimitations for ideal weight and height. Both the patient's weight and height are within normal limits, even though between the age of 2.2 and the age of 2.11 it was noticed a sudden weight increase, from the minimum limit to the maximum one.

Biological age versus chronological age

It is noticed that there is a major discrepancy between the chronological age and the biological one, both at the beginning of personalized physical therapy sessions, as well as in present.

However, compared to the biological age from 7 months ago, there is amelioration, considering that the patient evolved during this period from the point of view of her biological age with approximately 2 months.

Tabel 1: Chronological age versus biological age, at the beginning of physical therapy sessions and in present

Date	Chronological age + characteristics	Biological age + argumentation
05.11.2013	*** 4 years and 2 months – at 4 years old, the child goes down the stairs by alternating her legs; they can stay in balance on one leg for 4-8 seconds. It is inherent that she acquired all the other neuro-motor components.	*** 2 months – she can support her head a little from ventral decubitus position. She does not support her head in sitting position without considerable efforts. She is alert to sounds, she smiles, she prattles and laughs. Sometimes she follows moving objects.
21.06.2014	*** 4 years and 10 months – at 5 years old, the child stands on one leg for minimum 8 seconds; they walk on tip toes long distances; they are ready to use and develop their motor skills for more complicated actions: writing, drawing, sewing, singing.	*** 4-5 months – she can support herself on forearms and elbows. She raises her head and bends the thorax. Suspended, she brings her head in the superior plan. She has control over her head in twisting movements. She responds to a smile by smiling. She prattles. She laughs loudly. She turns her head to the direction from where she is called.

GMFCS – 5th degree. The severe motor insufficiency limits willing control of movements and the ability to hold the head and control the trunk. All motor functions are limited. The restriction of the sitting function and standing position cannot be fully compensated by using adjustment equipment and assistance technologies. The child with 5th degree gross motor abilities cannot walk alone, therefore they are transported. (Robert Palisano and co., 2007)

Postural assessment. The spinal curves have never been formed, considering that the patient has never taken independently the sitting and orthostatic positions. The back muscles have also been, and still are, hypotonic. Still, the patient has been assessed with the scoliometer from sitting and bent forward positions (supported, so as she would not fall laterally) and it has been noticed that she presents a 5° thoracic scoliosis. From sitting position, the patient takes a position of kyphosis in D. She also presents plantar flexion at both feet, caused by the retraction of the Achillean tendon.

Effects of Bowen therapy. As effects of Bowman therapy, the family noticed that the patient would sleep better during the night and, besides this aspect, it was noticed a reduction of grinding her teeth together and of her tick of continuously pulling out her tongue.

Effects of using the Dondolino device. The Dondolino device was used before and during the period of undergoing Bowman therapy and no effect was noticed.

DISCUSSIONS

Aspects concerning growth in height and weight. Growth disorders are a characteristic of children with Rett's syndrome, yet, there are no specific growth graphics. D.C. Tarquinio and co. (2010) determined in their paper work referring to growth disorders in Rett's syndrome that the growth disorders in head circumference begin at 1 month, those in weight at 6 months and in height at 17 months. The BMI was similar to that of normal people. They also noticed that many co-morbid disorders had a strong impact upon the growth of children suffering of Rett's syndrome, such as: ENT-pharynx and gastro-intestinal disorders, scoliosis, seizures and osteopenia.

Kathleen J. Motil and co. (2013) determined in their paper work referring to prevalence of gastro-intestinal disorders, that in 47% of the cases, deficit or excess weight occurs and at 45% of the cases, growth deficit occurs. In the case of the studied subject, although the patient suffers of dysfunctions such as: mastication disorders, gastrointestinal dysfunctions, scoliosis, comitial seizures, she is within the normal weight and height limits.

However, considering her weight, it seems to evolve towards an increase above the normal limit as until the age of 2.2, she was at the lower limit, from the age of 3, her weight suddenly increased

until almost the upper limit and it continues to increase constantly so, by the age of 5, it will reach the upper limit.

It is possible that, since her scoliosis is not very severe (4°), it does not influence the evolution of her height until present.

Scoliosis related aspects. Alan K. Percy and co. (2010), in their study on a lot of 544 participants with Rett's syndrome, referring to the scoliotic profile, noticed that 53% of them had scoliosis. The average age of subjects with scoliosis is 15 and of those without scoliosis is 6. In these participants, the presence of scoliosis was associated with: reduced score of motor behavior analysis, late motor acquisitions, loss or absence of walking function and constipation.

The results of this study are similar to the results of the present study as the patient is approximately 5 years old and she presents only a mild scoliosis (4°). Based on the study results, we can say that in time the condition can become more severe unless kinetic means are applied.

Gastro-intestinal problems related aspects. Kathleen J. Motil and co. (2013) initiated a national study to determine the prevalence of most frequent gastro-intestinal and nutritional disorders of patients suffering of Rett's syndrome based on parental report, the results being correlated with age. They elaborated a questionnaire referring to symptoms, diagnosis, diagnosing tests and therapeutic interventions concerning gastro-intestinal and nutritional problems in Rett's syndrome. The questionnaire was distributed to 1666 families, out of which 983 identified symptoms and diagnoses associated to gastro-intestinal des-motility (92%), mastication and deglutition difficulties (81%), weight deficit or excess in 47% of the cases, growth deficit in 45% of the cases.

In the case of the studied subject, the gastro-intestinal problems have manifested themselves by the presence of constipation ever since the first year of life until present time. Stool occurs once in 2-3 days, most of the times with the help of medication or by rectal stimulation with the thermometer. After the physical therapy sessions from the last half of year, the parents noticed that defecation required less medication intervention and it took place without considerable efforts from the subject.

Sleeping problems related aspect. The sleeping problems are frequent in children with Rett's syndrome, yet there are few research studies concerning their prevalence. Deidra Young and co. (2008) initiated a prevalence study on the Australian population which included 300 subjects with Rett's syndrome, aged between 2 and 29, born between 1976 and 1993, referring to functional abilities, sleeping behavior and patterns, medical and geno-typical conditions. The purpose of this study was to investigate the type and frequency of sleeping problems related to age and genetic mutation. It was also studied the change of sleeping pattern in time.

They noticed that the sleeping problems occurred in 80% of the studied patients. The most frequent sleeping disorders were reported as being grinding the teeth together, followed by screaming and seizures, frequent sleeping episodes during daytime (77%) and waking up during the night (especially in the 0-7 year-old group). (Deidra Young and co., 2008). According to the facts noticed in the mentioned study, the subject in discussion also presents sleeping disorders: frequent sleeping episodes during daytime (2-3, for approximately 1-2 hours), waking up during the night and random seizure, which decreased when Depankin was administrated. When the patient benefited of Bowen therapy, the parents noticed the amelioration of sleeping disorders and the decrease of their frequency.

Effects of the personalized kinetic program. According to the initial assessment in November 2013, we noticed the following aspects: the patient was in the biological age of 2-3 months. He did not raise her head, from VD and LD she supported it for a very short period of time (approximately 2 seconds). She moved her upper and lower limbs but in a chaotic manner, without a definite purpose. She did not role on either side. She could not sit and, if positioned like that, she had to be supported both from lateral and from the front and behind concomitantly. Her feet were spastic in permanent position of flexed toes, inversion and plantar flexion. Her anterior tibial was permanently elongated and her Achillean tendon, permanently shortened.

Her hands and feet were permanently cold (possible slow blood flow). In time, after approximately one month with 2 sessions per week, it was noticed that she held her head much better from sitting position, she could stay supported on her forearms with raised head and she rolled on both sides with help in initializing the motion. After mid-February, the following progresses were noticed: she raised her bottom – approximately 20 partial raises, she held her head much better and for longer,

she rolled alone to the right and with help to the left; on the ball she had shorter breaks between head raises. In April, 2014 it was noticed that, pulled by her UL on diagonal, she did not initiate the rolling motion, but she supported herself on the elbow of her other UL, later on the hand and, helped out, she reaches the sitting position.

However, unfortunately, these progresses are neither permanent nor on long term. When she becomes ill, the patient takes long breaks between sessions (even 2 weeks), fact which constantly determines ups and downs in her neuro-motor evolution.

Presently, the improvements noticed in time have been maintained. In the following images it can be noticed the way in which the patient stays in the low doll and sitting postures.

Even though these positions are maintained for a short period of time (under 1 minute), it is still a progress, considering that at the beginning she would not reach these performances at all. At present, the patient does not need stimulation at shoulder level to raise her head laterally. She does it alone, as soon as she is positioned in LD.

It is noticed an evolution of her support on forearms from VD, since March, when the patient held both her forearms at her chest, unlike June, when she started to take out one forearm at a time from underneath her.

CONCLUSIONS

In conclusion, according to the results, even if the patient is between normal limits regarding her weight and height, she fits the typical framework of the child with Rett's syndrome regarding the mastication disorders, gastro-intestinal dysfunctions, scoliosis, comitial seizures and sleeping disorders. Although her weight is within normal limits, it can increase over this limit. It can also be said that, in time, it may become more severe if kinetic means are not applied.

Concerning the secondary dysfunctions, improvements were noticed as a result of practicing various therapies: the seizures were considerably reduced as frequency and intensity after the treatment with medication; the sleeping disorders and grinding the teeth ameliorated when practicing the Bowman therapy; after the oxygen therapy, it was noticed an improvement of the patient's attention; after the physical therapy sessions from the last half of year, the parents noticed that defecation required less intervention with medicines and took place without considerable efforts from the subject. These improvements were completely lost however, as a consequence of seizures onset. After the physical therapy sessions combined with Bowman therapy, the patient evolved from the point of view of her biological age with approximately 2 months (from 2 months to 4-5 months).

In conclusion, it can be stated that a personalized physical therapy program, combined with Bowman therapy, can have beneficial effects upon the neuro-motor and functional development of the child suffering of Rett's syndrome.

CONFLICT OF INTEREST

 There aren't any.

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