CARACTERISICI ŞI LIMITE ALE KINETOTERAPIEI ÎN RECUPERAREA BOLILOR GENETICE – STUDIU DE CAZ

CHARACTERISTICS AND LIMITS OF PHYSICAL THERAPY IN REHABILITATION OF GENETIC DISORDER - CASE REPORT

Adina Mitrică, Ada Petrache[9], Carmen Avram, Felicia Cioara[10]

Key words: **physical therapy, rehabilitation, Silver Russel Syndrome**

Abstract. Some authors separate the Russell and Silver syndromes as independent entities, however, Russell's and Silver's are now considered the same entity. This is a rare syndrome, more than 400 cases have been reported. The first reports were in children with characteristic face, low birth weight, asymmetry, and growth retardation. Growth failure is the primary abnormality. Patients typically present with intrauterine growth retardation, difficulty feeding, failure to thrive, or postnatal growth retardation. The clinical features are more evident in infants or young children.

Methods. We present a little girl age 1 year and 7 months, admitted in our hospital for functional rehabilitation having moderat to severe neuromotor retardation, associated with severe malnutrition, low weight, congenital heart malformation, gastroesophagian reflux. We evaluated the child using articular and muscular testing, GMFM scale. We applied a kinetotherapy program in order to achieve motor skills according to chronological age.

Results. We followed up the child over one year. Rehabilitation treatment improved muscle tone, motor skills, posture and spine alignement.

Conclusions. An early intervention program, in this particular case is beneficial.

Also a specific physicaltherapy program at home followed systematic improved growth and development in this case.

Cuvinte cheie: kinetoterapie, recuperare, Sindrom Silver Russel

Rezumat. Unii autori separa sindromurile Russell si Silver drept entitati diferite, desi, acum ele sunt considerate aceeasi entitate. Acesta este un sondrom rar, s-au reportat mai mult de 400 de cazuri.

Falimentul creşterii în perioada postnatală ridică numeroase probleme de diagnostic şi tratament. Prima anormalitate este esecul cresterii. Sindromul Silver Russel este caracterizat prin dismorfism craniofacial, greutate mică la naştere şi retard al creşterii în greutate şi înălţime si sunt mai evidente la sugari si copii mici.

Material şi metodă. Prezentăm cazul unei fetiţe în vârstă de 1,7 ani, care se interneaza pentru tratament de recuperare functionala prezentind malnutriţie protein energetică cu hipotrofie staturoponderala, retard neuromotor moderat sever, asociat prezintă defect septal atrial, boală de reflux gastroesofagian. Am evaluat copilul folosind testare articulara si musculara, pe scara GMFM. Am aplicat un program de kinetoterapie pentru a obtine abilitati motorii potrivite varstei cronologice.

Rezultate. Am urmărit obţinerea achiziţiilor motorii, corespunzătoare dezvoltării pe etapă de vârstă, pe o perioadă de aproximativ un an. Tratamentul de recuperare este efectuat sistematic pînă la ameliorarea indicilor funcţionali.

Concluzii. Studiul acestui caz aduce un argument în plus pentru tratamentul de recuperare instituit precoce, individualizat şi pe o perioadă îndelungată de timp.

[9] PT, „Felix Spa" Medical Rehabilitation Clinic Hospital, „1 Mai" Children Department
[10] MD, „Felix Spa" Medical Rehabilitation Clinic Hospital, „1 Mai" Children Department

Introduction

Silver-Russell syndrome (SRS) originally was described by Silver and colleagues in 1953 and, soon afterwards, by Russell in 1954. The first reports were in children with characteristic facies, low birthweight, asymmetry, and growth retardation. (12)

Over the past several years, more than 400 patients have been described, with phenotypes ranging from mild to classic. Estimates of incidence range from as high as 1 in 3,000 to as low as 1 in 100,000. No sex preponderance, the male-to-female ratio is equal.(1)

SRS is both clinically and genetically a heterogeneous disorder, and the basic underlying defect is not known. SRS usually occurs sporadically and its etiology is not identified in most cases. A variety of molecular defects have been reported, some patients have maternal uniparental disomy of chromosome 7, with the possibility of imprinting (inheriting 2 copies of maternal chromosome 7, with no paternal contribution).(4)

As seen in literature SRS is a syndrome of intra-uterine dwarfism with low birth weight characterized by short stature, skeletal asymmetry, relatively large skull with triangular shaped head, small incurved little fingers, characteristic facies, precocious sexual development.(3) Renal and/or ureteral defects and mental retardation may occur in some cases.(9) Café au lait macules are present in nearly half the cases. Nephroblastoma can be a complication. The mode of inheritance is unknown.(8)

Pathophysiology

Growth failure is the primary abnormality. Patients typically present with intrauterine growth retardation, difficulty feeding, failure to thrive, or postnatal growth retardation.(7) Facial dysmorphism exists, with small triangular face and normal head circumference. Because length usually is less than normal, the head appears disproportionately large. Intelligence may be normal, or the patient may have a learning disability.(11)

Infants have failure to thrive, feeding difficulties, and fasting hypoglycemia.

Treatment

Medical Care is most of all oriented on growth and development. An early intervention program, including special education programs, physical therapy, and occupational therapy is beneficial. Special education courses are needed when the child is older. Also, significant effort must be undertaken to optimize caloric intake.

Surgical care must be consider in early childhood, as enteral feeding if the patient does not tolerate oral feeding and has severe failure to thrive. As this kind of children raise up a lot of problems during his care, a complexe team of specialists will be needed during the treatment, as it follows: clinical geneticist, a gastroenterologist or nutritionist, pediatric endocrinologist, rehabilitation physician, physical therapist, logopedist, occupational therapist, nutritionist, paediatrics specialist, psychologist and family, provides the favorable evolution.

The prognosis is relatively good. Some patients may have a learning disability.

Special attention must be on patient education, especially on educate the family regarding the growth and development of these children. Genetic counseling is important, because most studies demonstrate that the siblings of children with SRS do not have the disorder.(5) Therefore, risk for recurrence is expected to be minimal. In some of the cases seen to date, the children of patients with SRS do not have the disorder.

Researching means and methods

The researching methods used by the authors were:

the method of bibliographic research

the method of clinical evaluation

a) the method of case investigation
b) the method of statistic examination
c) data gathering

The fundamental means of physical therapy used by the authors in the rehabilitation of Silver- Russel syndrome child were:

a) physical exercise
b) massage
c) The helping means of physical therapy carried out for this issue were represented by the physical treatments which are specific to the department:
   a) thermo-therapy
   b) electrotherapy
   c) hydrotherapy
   d) occupational therapy
   e) adjusted physical activities

The means related to physical therapy, which the patient beneficiated by, were:

a) natural factors: water, air and sun
b) hygienic factors and hyper-caloric food

The techniques and the methods, specific for physical therapy, were:

a) basic kinetology techniques: - akinetic techniques
   - kinetic techniques
b) stretch-reflex
c) techniques of proprioceptive neuromuscular facilitation (PNF):

- PNF general techniques
- PNF specific techniques

   a) mobility techniques
   b) stability techniques
   c) controlled mobility techniques
   d) ability techniques

d) physical-therapy methods: Bobath method(cordun2)

Physical therapy applied in the rehabilitation process of Silver- Russel syndrome child

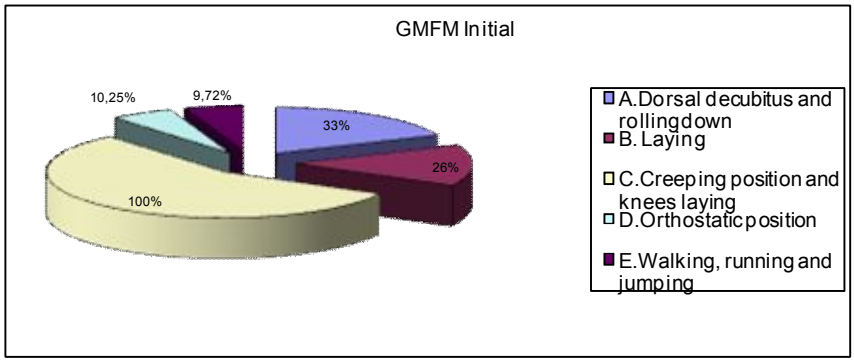
Initial assessment:

- Staturo-ponderal hypotrophy, 67cm, 5200g, cranial perimeter 47cm (ideal for this age; 81,5cm, 11kg) harmonious,
- Characteristic facies, triangular face, prominent frontal bumps, fontanel 2/3 cm, pigmented teguments
- Spine in axle, axial hypotonicity, dorso-lumbar kyphosis increased in sitting position, postured in orthostatic position hyperlordosis
- Upper lambs, lower lambs hypotonicity, symmetrical muscular hypotrophy, foot with plano-valgus propping up
- She doesn't raise her head in dorsal decubitus position, upheld from the thoracic level by the physical therapist.
- She can't get to staying position.
- She can't get the orthostatic position, independently.
- Walking: not achieved at first visit, then only with bilateral upholding, she steps with a large supporting base, dysmetric, stability, equilibrium disorders.

Table no. 1: Initial assessment – GMFM record

No.	**GMFM**
	Initially
A. Dorsal decubitus and rolling down	33%
B. Laying	26%
C. Creeping position and knees laying	100%
D. Orthostatic position	10.25%
E. Walking, running and jumping	9.72%

Chart no.1 Initial assessment - GMFM record



Goals:

A. Getting the sitting, orthostatic, walking position
B. Improving the postural control
C. Improving the tonus muscular disorders (hypotonicity), the statokinetic disorder
D. Getting accustomed to strategies of equilibrium control (the strategy of the ankles, knees, hips) and to controlled falls
E. Toughening the muscular groups involved in the breathing process
F. Increasing the withstanding of the effort with the supervision of the biologic test of glycemia
G. Forming the reflex of correct corporal posture in static/ dynamic instance (s10)

A. The local static reaction represented a mechanism of maintaining the orthostatic position as a response to the action of the gravitational forces. All corporal connections were modified:

- The viewpoint on the environment has changed.
- The modifying of the back position has showed up.
- The insteps supported the pressure of the whole body and this fact led to new sensations coming out
- A new centre of mass showed up, precisely placed inside the supporting polygon

Reaching these transformations is very stressing and during the first stage it is associated to balancing reactions which are obvious especially while the child is walking.

a b

Fig. 1 (a,b) Getting the orthostatic posture by sitting

Getting the orthostatic posture was done in laying position, in knees position, in quadruped position, with or without help or uphold. All these were possible after the child learnt and consolidated the "ready to jump" protection reflex. The child stood up from the fore-mentioned position, experiencing "the elephant position''.

B. By postural control, the balance and the stability of the whole body, but of its segment too are maintained. The static balance and the postural control are got by the contraction of some muscular groups of the trunk related to the head position in space. The particularities of this issue materialised in the disproportion between head and trunk required an implementation of a kinetic program, rigorously attended for a long period, and the addition of some exercises to train the postural usage.

C. The permanent stimulation of motility, thinking and speaking used in child's making more active and the request of actions naming improve the perfecting of psycho-motility and of speaking.

The first stage is that of sensorial-moving intelligence (0-2), when the intelligence comes from the child's affective action with objects (purely practical intelligence), the action being the child's primary form of thinking. The principal instrument of adapting the child to reality is the initial sensorial-moving scheme and the action is the way of solving the adapting problems. It was essential, during the treatment, for the patient to receive as many sensations as possible, concerning the tonus, the posture and the normal moving.

Fig. 2 Educarea ridicarii in asezat prin lateral cu sprijin pe coate si antebrat pe minge ajutat de kinetoterapeut

D. The treatment consisted of the training of "moving modalities". To fortify the musculature, neuro-proprioceptive facilitation techniques, stretch- reflex were used and the analysis and action principles in accordance with Bobath method were applied, the purpose being to increase muscular tonus. So, neuro-proprioceptive facilitation techniques used in research were: IL, ILO, CR, IZA, SR. To fortify the flexors head- neck, the Bobath ball was used in a large measure to facilitate the moving performance. In order to fortify the musculature of the lower limbs sand purses were used. Diverse sensorial systems were stimulated, activating the receptors from the level of the tegument, conjunctive, muscular and articular tissue, by manual contact, touching, traction, pressure, vibrations.(z6)

E. The child was taught to provide her body balance during the movements by muscular activations called posture adjustments. A difficult problem in the postural performances was represented by the coming out of a lack of balance, in the inactivated muscles in the moment of recovery. Four types of strategies were used and they developed in stages in accordance to the intensity of the unbalancing stimulus:

- Ankles strategy
- Hips strategy
- Suspension strategy
- Steps strategy

The automatic postural performances weren't challenged only in orthostatic position, released themselves as a typical effect of balancing in laying position too.

F. Training the breathing was essential and obligatory:

- training the costal breathing
- training the diaphragmatic breathing (we didn't insist because of lumbar hyperlordosis)
- control and coordination of breathing

Physical therapy, in the case of the children with breathing disorders, had in view: - to know air absorption (nose inspiring, mouth expiring)

- to re-balance the musculature involved in breathing.

Hydro-physical therapy was used with extern resistance. The special programs for children consisted of the correction of the pathological curvature of neck and head coordinated to breath, of shoulders and back, of dorsal and lumbar spine, of pelvis position and of mobility in quadruped posture, of the typical re-education of the diaphragm. As concerns the training of air absorption, the exercises had come true by:

Forming air bubble in water

The patient blows in a tube which is introduced in a bottle of water. She blows slowly and continuously so that an uninterrupted series of bubbles to form. Timing the duration of the expiring (fact that is offered by the production of bubbles) is a good test of the mobilizable volumes and of the flux resistances. The height of water in the bottle represents the interposed resistance, offering the opportunity to estimate expiratory force (passive or active in accordance to the way of executing the expiring). The consideration of this test in the evolution of this disease and of recovery assistance is satisfactory.

Increasing the inspiring ability (by means of a special device)

The patient is in laying position, introduces the apparatus pipe in her mouth and pushes, by inspiring, the ball of the device as high as possible, maintaining it. The device has various difficulty degrees in order to get an efficient inspiring. Timing the duration of the inspiring is a good test of breathing flux resistance.

Increasing the expiring capacity (by means of a ping-pong ball)

The patient is in laying position and she is asked to blow slowly and continuously the ping-pong ball which is on the table, in front of her, with two pillows laterally in order to prevent the ball falling after a profound inspiring. We can estimate the expiring capacity of the patient by seeing the length of the tape measure – from the moment of putting the ball on the table till its stop.

G. To obtain the stress withstanding, it is relevant to know which the starting level and the final one were and what physic training means, adapted to this pathology were used. Child's testing for stress was necessary in order to individualise the program of physical training and to determine the stress

tolerance. The used methods were: walking, walking on accidental ground, self-care activities, platforms mounting, the rolling carpet, running, occupational therapy. The endurance of physical effort was supervised by the means of the following clinical effects: the ones which indicated hypo-glycemia (drowsiness), tachycardia, pallor, cold sweat, vertigo, irascibility, periods without weight gain.

H. In order to correct the hyperlordosis the following hypotonic muscles were fortified:

- abdominal ones, especially the right abdominals
- external rotators of lambs
- quadriceps
- pre-cervical ones

Final assessment

- As concerns the staturo-ponderal issue the measures show a height of 80 cm and a weight of 7000 g
- The spine has physiologic curvatures
- The upper lambs present a normal muscular development and the patient can use them in order to get ADL
- The lower lambs present a muscular fortifying which permits the child to walk independently for long distances, to walk on accidental ground, to mount the platform, to bring down the slopes, to climb – to lower the stairs, to run
- In dorsal decubitus position, stabilizing the lower part of the body, she gets head – neck flexion with elbow support
- She obtains orthostatic posture independently
- She has balance reactions in all postures
- She can't get the laying position independently because of the disproportion between head and trunk

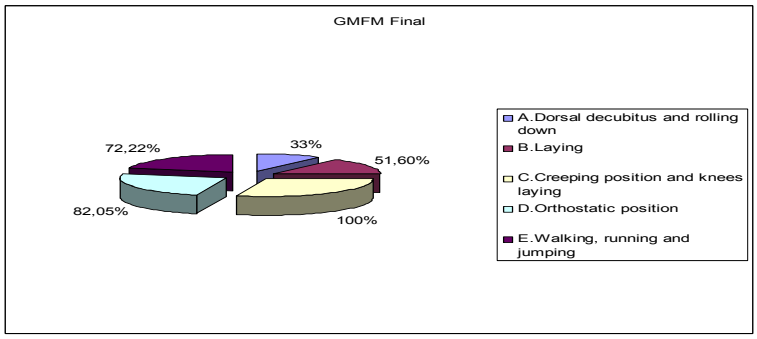
a b

Fig 3(a,b) The difference by initial assessment and final assessment in correct aligment of spine posture

Table no. 2: Final assessment – GMFM record

No.	GMFM Finally
A. Dorsal decubitus and rolling down	33%
B. Laying	51.6%
C. Creeping position and knees laying	100%
D. Orthostatic position	82.05%
E. Walking, running and jumping	72.22%

Chart no. 2 Final assessment – **GMFM** record



Conclusions and recommendations

- The precocious treatment for the children with genetic motor deficit influences the recovery and the following possibility of social integration decisively.
- The movement motivation is very important during the whole treatment.
- During the physical therapy programs, the child must receive as many, correct and close to normal sensorial information on posture and movement as possible.
- Efficiency and strictness of the programs concerning the personal hygiene, the nourishment (it is indicated not to eat an hour before and after the kinetic session) must be observed, and in this case physical therapy isn't suitable if it isn't associated to a hyper-caloric diet.
- A special attention must be paid to corporal scheme development, the child being able to point correctly, when requested verbally, the principal parts of the body, face, trunk, but also the fundamental segments of extremities.
- Physical therapy will aim to achieve the psycho-moving aptitudes especially the static function, the dynamic one, the global coordination, the manual abilities, the trunk suppleness, the speed of arms movement, the force of the lower limbs.
- Getting a correct alignment of the column has constituted the key of the other objectives materialisation successfully and soon it will facilitate the child's integration in a normal kindergarten.

Fig 4. The difference by children waist with SRS and Bobath ball with 65 diameter

Fig 5 The children height with SRS

Bibliography

1. Puiu, Maria, Principalele 100 de boli rare, *Orizonturi academice*, 2007, Timişoara
2. Cordun, Mariana, Kinetologie medicala, *Editura Axa,* Bucuresti, 1999

3. Duncan PA, Hall JG, Shapiro LR, Vibert BK. Three-generation dominant transmission of the Silver-Russell syndrome. *Am J Med Genet*. Feb 1990;35(2):245-50. [Medline].
4. Eggermann T, Wollmann HA, Kuner R, et al. Molecular studies in 37 Silver-Russell syndrome patients: frequency and etiology of uniparental disomy. *Hum Genet*. Sep 1997;100(3-4):415-9. [Medline].
5. Eggermann T, Meyer E, Ranke MB, et al. Diagnostic proceeding in Silver-Russell syndrome. *Mol Diagn*. 2005;9(4):205-9. [Medline].
6. Pasztay, Zoltan – Kinetoterapia in neuropediatrie*, Editura Arionda*, Oradea, 2004
7. Kotzot D, Schmitt S, Bernasconi F, et al. Uniparental disomy 7 in Silver-Russell syndrome and primordial growth retardation. *Hum Mol Genet.* Apr 1995;4(4):583-7. [Medline].
8. Langlois S, Yong SL, Wilson RD, et al. Prenatal and postnatal growth failure associated with maternal heterodisomy for chromosome 7. *J Med Genet*. Nov 1995;32:871-5. [Medline].
9. Moore GE, Abu-Amero S, Wakeling E, et al. The search for the gene for Silver-Russell syndrome. *Acta Paediatr Suppl*. Dec 1999;433:42-8. [Medline].
10. Sbenghe, Tudor , Russell A. A syndrome of "intrauterine dwarfism" recognisable at birth with craniofacial dysostosis, disproportionately short arms and other abnormalities (5 examples). *Proc Royal Soc Med.* 1954;47:1040-4. Kinetoterapia profilactica, terapeutica si de recuperare, *editura Medicala,* Bucuresti, 1987
11. Tanner JM, Lejarraga H, Cameron N. The natural history of the Silver-Russell syndrome: a longitudinal study of thirty-nine cases. *Pediatr Res*. Aug 1975;9(8):611-23. [Medline].