# MEDICAL REHABILITATION IN HEREDITARY NEUROPATHIES

## MARIANA CEVEI<sup>1</sup>, DORINA STOICĂNESCU<sup>2</sup>, RAMONA SUCIU<sup>3</sup>

<sup>1,3</sup>University of Oradea, <sup>2</sup> "Victor Babeş" University of Medicine and Pharmacy Timişoara

Keywords: hereditary neuropathies, medical rehabilitation, activity of daily living Abstract: Hereditary neuropathies represent a group of inherited disorders with progressive evolution that are typically classified in hereditary motor neuropathies, hereditary motor and sensory neuropathies, and hereditary sensory and autonomic neuropathies. The objective of this study was to highlight the role of medical rehabilitation, aiming to gain maximum of functional and occupational independence in patients with different types of hereditary neuropathies. This study was conducted on 143 cases diagnosed with hereditary neuropathies, admitted in the Medical Rehabilitation Clinical Hospital "Băile Felix", Romania. In adults, research focused on identifying the main procedures needed to increase functional independence and in children, research focused on the analysis in terms of acquiring walking independence with or without aids. Mean age of investigated children was 9.45 years, which pleads for even earlier start of medical rehabilitation. Monitoring the evolution of clinical and functional parameters revealed an improvement of activities of daily living after physical-kinetic therapy initiated early.

## INTRODUCTION

Hereditary neuropathies represent a group of inherited, chronic disorders with progressive evolution that are typically classified in the following types:(1)

- hereditary motor neuropathies
- hereditary motor and sensory neuropathies
- hereditary sensory and autonomic neuropathies

Hereditary motor neuropathies are hereditary disorders caused by degenerative lesions of anterior horn motor neuron, bulbar nuclei and/or corticospinal tracts.

Peripheral neuropathies represent a group of hereditary degenerative disorders, mainly affecting the peripheral nervous system. Hereditary motor and sensory neuropathies are characterized by progressive muscle weakness, pes cavus, depressed tendon reflexes, as well as sensorial affection in various degrees.(2) They can be classified according to their inheritance patterns, age of onset and clinical features. Family history, clinical assessment with particular attention to the type of affected nerve fibers, electrophysiological and pathological data enable the establishment of the type of neuropathy. In the investigated families, these data support the hypothesis that damage of axons causes neurological dysfunction and treatment is directed towards preventing degeneration of axons and their regeneration.

The inheritance can be autosomal dominant, autosomal recessive or X- linked recessive. Genetic counselling and establishment of the recurrence risk depend on the inheritance pattern of each case. Molecular genetic studies have had a major impact in understanding these groups of diseases, their classification is currently possible depending on the type of mutant gene and on the pathological protein that causes the disorder.(3)

Spinal muscular atrophies (SMA) are a group of neurodegenerative disorders characterized by loss of motor neurons in the anterior horn of the spinal cord. The most common form is proximal SMA, with autosomal recessive inheritance.(4) Clinical manifestations are represented by degeneration of spinal cord motor neurons, atrophy of skeletal muscles, muscle weakness and paralysis.(5) The disease is classified into four grades of severity (I-IV) based on age of onset and motor function achieved.(6)

Hereditary sensory and autonomic neuropathies are a group of conditions with very low incidences, genetically and clinically very heterogeneous, with different types of inheritance patterns. They are progressive sensory neuropathies with variable motor and autonomic involvement.(7)

Hereditary neurological diseases are relatively common in pediatric neurology. The onset age may largely vary, from prenatal life to late adolescence, but the initial symptoms of most of them occur during childhood and congregate in a certain age group.(8)

Late-onset forms of hereditary neuropathies were also described and genetic heterogeneity within them is likely.(9)

#### PURPOSE

The objective of the study was to highlight the role of medical rehabilitation, aiming to gain maximum of functional and occupational independence in patients with different types of hereditary neuropathies.

## MATERIALS AND METHODS

This study was conducted on 143 cases diagnosed with various forms of hereditary neuropathies. The objectives were different, depending on the age category that included cases which formed the study group, which is why we divided the study group into two groups: adults (over 19 years old) and children (1-18 years old).

Thus, batch I included 43 cases investigated during the period 01.01.2009- 27.10.2015 in the Medical Rehabilitation Clinical Hospital "Băile Felix", Romania, Department of Adults, and research focused on identifying the main procedures needed to increase functional independence – activities of daily living

<sup>&</sup>lt;sup>2</sup>Corresponding author: Dorina Stoicănescu, P-ţa. E. Murgu, Nr. 2, Timişoara, România, E-mail: dstoicanescu@yahoo.com, Phone: +40259 412834 Article received on 21.12.2015 and accepted for publication on 06.03.2016 ACTA MEDICA TRANSILVANICA March 2016;21(1):55-58

1

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Study batch II included 100 patients investigated and diagnosed with hereditary neuropathy admitted to the Medical Rehabilitation Clinical Hospital Băile Felix, Romania, Clinical Department of Rehabilitation for Children, and research focused on the analysis in terms of acquiring walking independence with or without aids.

## Means and methods of kinetic rehabilitation:

1. - posture in functional position for shoulderelbow-fist using ortheses. Mobile orthoses, simple splints, adhesive tapes can be used.

- alternating postures, free, active, carried out by the patient with the healthy extremity,

2. Prevention of joint mobility limitations: passive motion, posture with dynamic ortheses in a  $90^{\circ}$  angle of the foot. If the injury is definitive early orthosis is indicated.

3. Increasing paralyzed muscles strength using analytical exercises for affected muscles; rapid stretching, applying PNF techniques (proprioceptive neuromuscular facilitation stretching) and facilitators - light touch with ice, vibration; Kabat facilitation schemes: flexion and extension diagonals; walking exercises and knees crawling, walking between parallel bars and on steps.

- Prolonged passive stretching on the antagonistic muscles, massage and local heat applications to combat contractures;

- Electrotherapy: use of excito-motor currents (exponential current);

- Passive stretching manoeuvres (stretch-reflex) repeated until muscle contraction is obtained (usually 4-5 stretchings);

- Passive motion, passive-active in all joints of upper extremities with short stretches at the end of the movement (fingers, fist, elbow, shoulder, scapula), in all plans and normal axes, all amplitude, to maintain kinesthetic image, biofeedback;

- "Imaginative" exercises in which the patient imagines, focusing on a movement that can not be executed due to paralysis;

- Hydrokinetotherapy - passive and autopasive mobilization in warm water (for pain relief, relaxation of antagonist, facilitating agonist);

- Facilitation schemes, Kabat method (flexion and extension diagonals for upper limbs applied depending on the type of lesion), initially asymmetric and symmetric unilateral then bilateral.

- Facilitators and PNF techniques, in particular rapid stretches (stretch reflex), performed either analytically or in motor integration schemes (Kabat diagonals);

- Active exercises throughout the range of movement, isometric exercises, active exercises with resistance;

- Sensitivity rehabilitation, in order: pressure and pain, proprioception, kinesthetic, thermal sensitivity (cold and then hot), discrimination of two points, stereognosis.

- Elevated posture, orthosis, Moberg gymnastics for the upper extremities, cryotherapy, massage, veno-lymphatic drainage, hydrotherapy (whirlpool-type bath), alternating baths, electrotherapy, local and general hygiene, combating vasculartrophic disorders.

- Occupational therapy exercises.

#### RESULTS

During 01.01.2009- 27.10.2015 in the Medical Rehabilitation Clinical Hospital "Băile Felix" Romania, a total number of 143 cases with different types of hereditary neuropathies were admitted, for medical rehabilitation. Distribution of cases according to gender did not reveal significant differences between genders, there were 71 men (49.6%) and 72 women (50.35%).

Mean number of admissions:  $2.87\pm3.43$ , with ranges between 1-20 admissions.

These 143 cases were divided into two groups, according to their age. The first group (batch I) included 43 adults and batch II was formed by 100 children.

Mean age of adult cases from batch I was 41.51 years $\pm 17.61$  years, ranges between 19 and 79 years (table no.1, figure no. 1).

Mean age of children from batch II was 9.45 years  $\pm 5.18$  years, ranging between 1.4 months and 18 years (table no. 2, figure no. 2).

Figure no. 1. Distribution of cases according to age in batch



Table no. 1. Distribution of cases according to age groups in batch I

Age groups (years)	No. of cases	% of cases
under 5	32	22.37
6-10	21	14.68
11-15	26	18.18
16-18	21	14.68

Figure no. 2. Distribution of cases according to age in batch



Table no. 2. Distribution of cases according to age groups in batch II

A	ge groups (years)	No. of cases	% of cases
	19-25	10	7
	26-30	5	3.5
	31-35	4	2.79
	36-40	4	2.79
	41-45	4	2.79

AMT, vol. 21, no. 1, 2016, p. 56

46-50	4	2.79
51-60	6	4.19
Over 60	8	5.59

Distribution of cases according to the diagnosis revealed the following data:

1. Hereditary cerebellar ataxia was diagnosed in 16 cases out of 143 cases (11.18%), 12 women and 4 men (table no. 3)

Table no.	3.	Distribution	of	cases	with	hereditary	cerebellar
ataxia							

	Number of	Gender	
Diagnosis	cases % of total cases	Males	Females
Friedreich's ataxia	9 (6.29)	1	8
Late onset cerebellar ataxia	1 (0.69)	0	1
Cerebellar ataxia with DNA repair defect	1 (0.69)	1	0
Congenital disorder of glycosylation	2 (1.39)	1	1
Unspecified ataxia	3 (2.09)	1	2

- 2. Hereditary motor neuropathies associated with CNS involvement was diagnosed in 5 cases out of 143 cases (3.4%) and according to the gender, there were 4 women and 1 man.
- 3. Hereditary motor and sensory neuropathies were diagnosed in 34 cases/143 cases (23.77%), 13 women and 21 men (table no. 4).

Table no. 4. Distribution of cases with hereditary motor and sensory neuropathies

Disensoia	Number of cases	Gender		
Diagnosis	% of total cases	Males	Females	
Charcot - Marie-	24 (16.78)	15	9	
Tooth syndrome				
Dejerine-Sottas	10 (6.99)	6	4	
syndrome				

 Spinal muscular atrophies (SMA) were diagnosed in 88 cases out of 143 cases (61.53%): 43 women and 45 men (table no. 5).

Table no. 5. Distribution of cases with spinal muscular atrophies (SMA)

Diagnosis	Number of	Gender	
	cases, % of total cases	Males	Females
SMA TYPE I (Werdning- Hoffman)	12 (8.39)	4	8
SMA TYPE II	32 (22.37)	18	14
SMA TYPE III	22 (15.38)	11	11
Scapuloperoneal SMA	22 (15.38)	12	10

Evaluation of the ability to perform activities of daily living, through observation, during the occupational therapy program conducted during hospitalization, proved its obvious improvements, especially the self-care section, by acquiring a certain degree of functional independence.

#### DISCUSSIONS

Inherited neuropathies comprise a group of neurologic diseases with clinical and genetic heterogeneity. The diagnosis will be established after complete neuromuscular evaluation, nerve conduction studies, laboratory testing, histopathologic examination and genetic testing. Treatment approaches are in the majority of cases only supportive.(10-12)

Hereditary disorders that occur due to mutations at different loci may have similar phenotypes. In these situations genetic risk depends on the type of inheritance for each of these mutations. This finding has practical implications in the genetic counselling of affected individual or their families. Genetic counselling and risk recurrence assessment of hereditary neuropathies must be individualized, depending on the inheritance pattern.

Hereditary neuropathies can not be cured yet, thus medical rehabilitation is very important. Physical therapy involves proprioceptive training, balance exercises, stabilization techniques for both upper and lower extremities, vestibular exercises for accomplishing functional improvement in cases with ataxia and where necessary supportive devices may be used.(13)

Functional evaluation requires compliance from the patient and patience from the physiotherapist. The focus is both an analytical assessment (goniometry, muscle testing) and on those functions and global movements, activities of daily living of the segment or extremity whose innervation is provided by the peripheral nervous structure of interested, assessing prehension. This establishes functional diagnosis.

Kinetic treatment will be phased: initially surgical intervention of the affected segment will take place and this will be immobilized in the functional position. Kinetic treatment targets the unaffected segment.

The second stage will represent rehabilitation itself, the duration of which can vary from 1 to 3 months until the musclea reach muscle force 1 (F1). Stage III begins when the muscles have reached F1 and reach force F3. Stage IV is the stage at which muscles fully recover from F3-F5 muscular force.(14)

The objectives of kinetic treatment will be tailored according to specific paresis or paralysis, step of treatment and recovery progress.(15)

The objectives of kinetic therapy are very complex: 1. Avoiding, correcting the appearance of deformities and vicious attitudes

- 2. Avoid paralyzed muscle atrophy
- 3. Maintaining joint mobility
- 4. Re-education of paralyzed muscles
- 5. Increase function of healthy muscle fibers
- 6. Regaining coordination of movements, functionality and ability
- 7. Education reeducation of sensitivity
- 8. Preventing and treating trophic disorders

Preventing pressure sores, bladder rehabilitation and prevention of urinary tract infections are also important.

In spinal muscular atrophies, the field of translational research is active and clinical trials are ongoing. Improving life expectancy and their quality of life are important targets in the care of these patients.(16)

#### CONCLUSIONS

- 1. Physical-kinetic program, occupational therapy, psychotherapy applied to patients with hereditary neuropathies are therapeutic means needed to achieve the targets.
- 2. Improvement of the ability to perform activities of daily living by acquiring a certain degree of functional independence was observed in all investigated cases.
- 3. Performing consistently the rehabilitation procedures in specialized centers is the essential condition for obtaining functional performances.

AMT, vol. 21, no. 1, 2016, p. 57

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