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Original Article

The effects of rehabilitation on functional independence of Eastern Taiwanese children with rare or genetic diseases

*Yu-Chuan Lin[a], Chun-Ying Weng[b], Chung-Chao Liang[c], Shao-Yin Chu[b,d,e]**

[a]Department of Nursing, Tzu Chi University, Hualien, Taiwan, [b]Department of Medical Genetics, Hualien Tzu Chi Hospital, Buddhist Tzu Chi Medical Foundation, Hualien, Taiwan, [c]Department of Rehabilitation Medicine, Hualien Tzu Chi Hospital, Buddhist Tzu Chi Medical Foundation, Hualien, Taiwan, [d]Department of Medical Education and Pediatrics, Hualien Tzu Chi Hospital, Buddhist Tzu Chi Medical Foundation, Hualien, Taiwan, [e]School of Medicine, Tzu Chi University, Hualien, Taiwan

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Abstract

Objectives: This study investigated the effects of outpatient rehabilitation therapy (RT) on the functional performance of children from Eastern Taiwan with rare or genetic diseases. **Materials and Methods:** This retrospective observational cohort study included 73 children from Eastern Taiwan who were affected with rare or genetic diseases, with an average age of 8.57 ± 5.33 years (47 boys and 26 girls). Each child received the goal-directed therapy known as outpatient RT, which was delivered by a multidisciplinary team of specialists. To assess the effectiveness of RT, the WeeFIM-C questionnaire data were collected and analyzed. **Results:** After receiving outpatient RT, most of the children only required low-to-moderate assistance with self-care tasks (4.36 ± 2.38), and they could perform mobility-related activities under supervision or independently (5.70 ± 2.29). Moreover, most only required minimal assistance with tasks related to cognitive functioning and tended to complete such tasks under supervision (4.97 ± 2.05). The functional performance was significantly different among three studied groups, in terms of self-care ($F_{[2, 68]}$ = 5.42, P < 0.007), mobility ($F_{[2, 68]}$ = 8.17, P < 0.001), cognitive functioning ($F_{[2, 68]}$ = 3.31, P < 0.042), and overall ($F_{[2, 68]}$ = 6.44, P < 0.003) functional performance. **Conclusion:** The results of this study demonstrated that the functional status was different among three studied groups in terms of self-care, mobility, and cognitive functioning after receiving outpatient RT.

Keywords: *Children with rare or genetic diseases, Rehabilitation therapy, WeeFIM-C*

Introduction

Providing a full range of care for children with rare or genetic diseases requires the long-term joint efforts of multidisciplinary professionals and a holistic approach. Such care is required to identify patients' functional status, which assists in individualizing care plans for disease control, personal development, family education, and community activities [1].

A retrospective cohort study revealed that patients' functional independence considerably improved during and after a multidisciplinary inpatient rehabilitation therapy (RT) program [2]. However, few studies have examined the effects of multidisciplinary RT on the functional independence of children with rare diseases. It is also unknown whether outpatient RT improves functional independence.

The present study examined the functional performance of daily living activities relating to self-care, mobility, and cognitive abilities among children with rare or genetic diseases in Eastern Taiwan and assessed whether outpatient RT implemented by a multidisciplinary team of professionals could improve functional performance.

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Materials and Methods

Study design

This was a retrospective observational cohort study.

Study cohort

The study included 73 children with rare or genetic diseases (47 boys and 26 girls) aged 1 to 21 years (mean age: 8.57 ± 5.33 years) who visited the Pediatric Genetic Clinic and Children' Rehabilitation Unit at Buddhist Tzu Chi Hospital between June 1994 and January 2015. Of these children, 43.8% (32/73) had diagnoses of rare diseases recognized

**Address for correspondence:* Dr. Shao-Yin Chu, Department of Medical Genetics, Hualien Tzu Chi Hospital, Buddhist Tzu Chi Medical Foundation, 707, Section 3, Chung-Yang Road, Hualien, Taiwan.
E-mail: chushaoyin@gmail.com

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by the Health Promotion Administration, Ministry of Health and Welfare [3], 32.9% (24/73) had chromosomal disorders, 57.5% (42/73) had single genetic disorders, 4.1% (3/73) had multiple congenital anomalies, and 5.5% (4/73) had psychomotor retardation. The cohort was stratified into three groups by the frequency of RT status: children who received RT no more than once per month (RT0), children who received RT once per week (RT1), and children who received RT at least twice per week (RT2).

Setting and intervention

Each child was prescribed patient-centered and goal-directed RT, which was implemented by a multidisciplinary team, including a rehabilitation physician, physical therapist, occupational therapist, speech therapist, social worker, nurse, dietician, and clinical psychologist. Relevant interventions included education, physical therapy, aquatic physical therapy, occupational therapy, strength and flexibility exercises, balance training, speech therapy, and nutrition counseling.

Measures

The effects of outpatient RT were assessed using the WeeFIM-C questionnaire, which was developed to assess functional independence in children aged 6 months to 7 years; however, it can also be extended for use among individuals with developmental disabilities aged up to 21 years [4]. The WeeFIM-C has 18 items categorized into the three domains of self-care, mobility, and cognition. The self-care domain has eight items for assessing eating, grooming, bathing, upper body dressing, lower body dressing, toileting, bladder function, and bowel consistency. The mobility domain has five items for assessing chair transfer, toilet transfer, tub transfer, walking, and stair climbing. The five items in the cognition domain assess comprehension, expression, social interaction, problem solving, and memory.

The WeeFIM-C uses a 7-point ordinal scoring scale. Each of the 18 items is rated with a score of 1 to 7. A score of 1 to 5 indicates that the child requires assistance with various daily activities related to the individual item. A score of 6 or 7 indicates that the child needs no help. Thus, the higher the score is, the more independent the child is. Before the start of the study, the research assistant received training in survey administration and data collection procedures. The consistency of the item ratings obtained by the research assistant was verified by the principal investigator (PI). Data were collected and entered into a study-specific database, which was then reviewed by the data manager for completeness and accuracy.

Statistical analysis

Descriptive statistics for the children's demographics and WeeFIM-C scores were calculated. The analyses for categorical variables (sex, family type, and primary caregiver) and continuous variables (age, duration of RT, length of follow-up, total number of RT sessions, and overall and domain WeeFIM-C scores) were calculated using Fisher's exact test and F tests, respectively. If the RT duration and age were significant in group comparisons, multivariate analysis of covariance (MANCOVA) was conducted for the WeeFIM-C sub-scores and total scores. Statistical significance was set at $P \leq 0.05$. All analyses were conducted using IBM SPSS Statistics for Windows, version 22.0 (IBM Corp, Armonk, NY, USA).

Ethical considerations

All data were collected as part of routine clinical assessments. The Institutional Review Board (IRB) of Buddhist Tzu Chi Hospital granted approval for the study (IRB103-128-A). The IRB also granted access to medical records.

Results

Demographic characteristics

Table 1 presents the demographic characteristics of the study cohort (n = 73) stratified into the three RT groups (RT0, RT1, and RT2) based on RT status. The ages ranged from 1 to 21 years (mean age: 8.57 ± 5.33 years) for the 47 boys and 26 girls. The length of RT ranged from 1 to 201 months (45.1 ± 50.0 months). The frequency

Table 1: Demographic and baseline characteristics of children with rare or genetic diseases (n=73) stratified into three groups (RT0, RT1 and RT2) based on rehabilitation therapy status

Variables	RT0 (n=8)	RT1 (n=31)	RT2 (n=34)	$F/\chi 2$	P
Age year	10.9±2.8	6.3±3.9	10.1±6.2	5.54	0.01
Male sex, n (%)	7 (87.5)	23 (74.2)	17 (50.0)	6.23	0.04
Medical condition*, n (%)					
Chromosomal disorder	3 (37.5)	14 (45.2)	7 (20.6)	8.65	0.19
Single genetic disorder	3 (37.5)	14 (45.2)	25 (73.5)		
Multiple congenital anomaly	1 (12.5)	1 (3.2)	1 (2.9)		
Psychomotor retardation	1 (12.5)	2 (6.5)	1 (2.9)		
Length of follow-up-months	112.5±53.8	64.1±43.1	104.5±68.9	4.79	0.01
Length of RT-months	12.4±35.0	62.2±46.4	37.1±51.3	4.32	0.02
Frequency of RT sessions	51.9±16.6	248.8±185.8	547.5±285.8	21.90	0.01
Nuclear family-total, n (%)	2 (25.0)	14 (45.2)	18 (52.9)	10.32	0.41
Mother caregiver-total, n (%)	4 (50.0)	25 (80.6)	29 (85.3)	31.23	0.01

*Chromosomal disorders include both numeric and structural anomalies; single gene disorders include inborn errors of metabolism and others; multiple congenital disorders include microcephaly and hemifacial microsomia; and the manifestations of psychomotor retardation include mental retardation, psychomotor retardation, severe psychomotor retardation, suspect mitochondrial disorder, hyperbilirubinemia with mild MR and suspect Citrullinemia. RT0: Children who received RT no more than once per month, RT1: Children who received RT once per week, RT2: Children who received RT at least twice per week and had met the RT goals. RT: Rehabilitation treatment, MR: Mitral regurgitation

of RT ranged from 8 to 1608 sessions (366.3 ± 290.3). The length of follow-up after diagnosis ranged from 7 to 258 months (88.2 ± 60.6 months). Thirty-four (46.6%) children were from nuclear families. The majority (79.5%) of primary caregivers were mothers. All variables except for family type and medical condition differed significantly among the three RT groups.

WeeFIM-C scores by rehabilitation therapy group

Table 2 and Figure 1 present the mean sub-scores for the self-care, mobility, and cognition domains as well as the overall WeeFIM-C scores for the three RT groups; the RT0 children scored the lowest in all of these categories. Overall, the patients exhibited the highest performance in the mobility domain; the majority of them were able to move independently.

Table 2: MANOVA results for WeeFIM-C scores by rehabilitation therapy group (*n*=73)

Group	RT0 (*n*=8)	RT1 (*n*=31)	RT2 (*n*=34)	*F*	*P*
Self-care score					
Range	8-56	8-56	9-56	4.57	0.014
Mean±SD	22.0±18.3	31.6±18.6	40.5±15.8		
Mobility score					
Range	5-35	5-35	5-35	8.22	0.001
Mean±SD	16.0±14.7	27.6±11.3	31.9±7.2		
Cognition score					
Range	5-35	5-35	8-35	4.35	0.017
Mean±SD	19.4±13.7	21.7±10.8	27.97±8.0		
Total score					
Range	18-124	18-126	31-126	6.48	0.003
Mean±SD	57.4±38.5	81.0±37.8	100.4±26.8		

Maximal possible scores: Total; 126, self-care; 56, mobility; 35 and cognition; 35. SD: Standard deviation, RT: Rehabilitation therapy

By contrast, most patients performed poorly on self-care tasks and tended to need minimal contact assistance or prompting, which assisted children in achieving performance levels of >75%. Therefore, children could perform cognitive tasks with minimal contact assistance to standby prompting, or they could perform them under supervision.

The radar map in [Figure 2] illustrates the scores for the 18 WeeFIM-C items for the three groups. The profile of the RT0 group for all 16 items tended to be the smallest, except for comprehension and expression items.

In the self-care domain, the children performed most favorably in eating, especially with supervision or moderate contact assistance (5.8 ± 1.9, 4.4 ± 2.6, and 3.4 ± 2.6 for RT1, RT0, and RT2, respectively). By contrast, the children performed least favorably in bathing and required additional assistance, ranging from minimal contact assistance to maximal contact assistance (4.1 ± 2.3, 2.7 ± 2.25, and 1.9 ± 2.1 for RT2, RT1, and RT0, respectively).

The children's mobility ranged from modified independence to requiring maximal assistance. The RT2 children typically move independently (6.4 ± 1.5). However, the RT0 children tended to need maximal contact assistance or prompting, with children exerting 25% to 49% of their efforts. The RT2, RT1, and RT0 children all performed most favorably when walking indoors and outdoors (6.8 ± 1.1, 5.7 ± 2.3, and 3.1 ± 2.7), whereas they did least favorably when ascending and descending stairs (5.9 ± 1.9, 5.3 ± 2.5, and 3.1 ± 2.7).

In the cognition domain, the RT2, RT1, and RT0 groups performed most favorably on memory tasks (5.7 ± 2.0, 5.1 ± 2.4, and 3.9 ± 2.9, respectively) and comprehension tasks (5.9 ± 1.7, 4.5 ± 2.5, and 4.8 ± 3.1, respectively) while

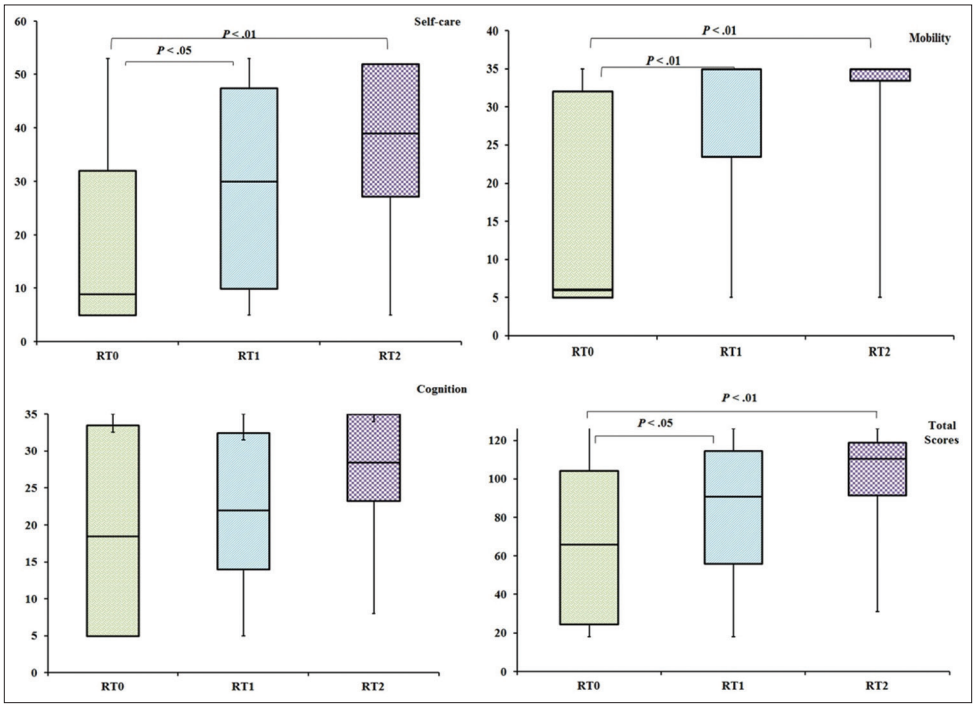

Figure 1: Self-care, mobility, and cognition domain scores and total WeeFIM-C scores for RT0, RT1, and RT2 after controlling for length of RT and age. RT: Rehabilitation therapy

performing least favorably on expression tasks (5.1 ± 2.4, 3.8 ± 2.6, and 4.5 ± 3.0, respectively).

Effects of outpatient rehabilitation therapy

MANCOVA was conducted to compare the effects of RT on children's functioning after RT duration and age were controlled for [Table 3]. The analysis revealed that RT significantly influenced the scores of the self-care, mobility, and cognition domains as well as the overall WeeFIM-C scores ($F_{[2, 68]} = 5.42$, $P < 0.007$; $F_{[2, 68]} = 8.17$, $P < 0.001$; $F_{[2, 68]} = 3.31$, $P < 0.042$; and $F_{[2,68]} = 6.44$, $P < 0.003$, respectively).

The effect of RT on children's self-care abilities was examined through a *post hoc* comparison with the Bonferroni method [Table 3 and Figure 1]. Significant differences between RT2 and RT0 ($P < 0.005$) and between RT1 and RT0 ($P < 0.02$) were observed, but the significant differences between RT2 and RT1were not observed. In short, the results demonstrated that children's self-care abilities were different among three studied groups, and the RT2 children possessed the strongest self-care abilities.

Significant mobility differences were also observed between the RT2 and RT0 groups ($P < 0.001$) and between the RT1 and RT0 groups [$P < 0.006$; Table 3 and Figure 1], but the significant differences between RT2 and RT1were not observed. This finding suggests that the RT2 children had the greatest mobility.

Although the results of MANOVA demonstrated significant differences between the three groups, the differences in cognitive scores between the RT2 and other groups (RT0 and RT1) were not significant [Table 3 and Figure 1]. Hence, the effects of RT on children's cognitive functioning were less effectively than it enhanced their self-care and mobility abilities.

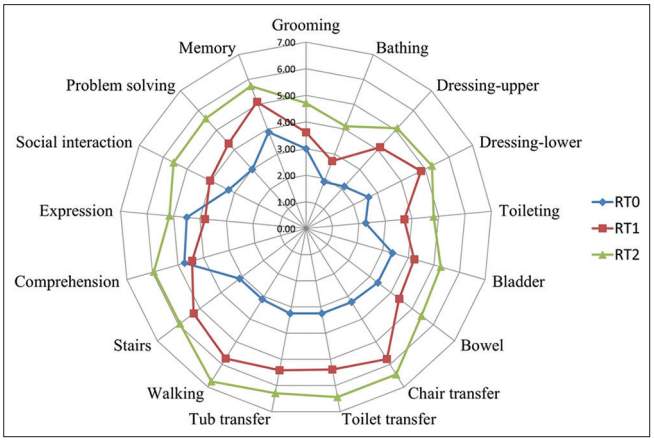

Figure 2: Radar map illustrating the WeeFIM-C scores for 18 functional performance items for RT0, RT1, and RT2. The profile of the RT0 group appears to be the smallest, except for comprehension and expression. RT: Rehabilitation therapy

Discussion

By comparing the WeeFIM-C scores of three RT groups, this study investigated the effects of outpatient RT delivered by multidisciplinary professionals on the functional performance - namely the self-care ability, mobility, and cognition - of children with rare and/or genetic diseases, and psychomotor retardation without a confirmatory diagnosis. As expected, the WeeFIM-C scores of the RT2 children significantly increased more than those of the RT0 and RT1 children [Table 3 and Figures 1, 2]. However, the *post hoc* analysis in Table 3 did not show any significant differences between RT2 and RT1 in WeeFIM-C scores, which is inconsistent with a previous study [5]. This may suggest that outpatient RT once per week benefits children with rare and/or genetic diseases, and psychomotor retardation without a confirmatory diagnosis just as much as outpatient RT twice per week does in terms of self-care ability, mobility, and cognition [2,5,6].

The differences in cognitive scores between the RT2 and other groups (RT0 and RT1) were not significant, which is consistent with a previous study [5]. This could be explained by these factors: a small sample size, heterogeneous groups, the complexity of pathophysiologic cognitive development, and the total number of RT sessions. Additionally, more RT sessions and time may be required in the cognition domain for children with rare or genetic diseases.

The contribution of this study is that we investigated how interdisciplinary teams of specialists conduct outpatient RT

Table 3: MANCOVA results for WeeFIM-C scores after controlling for length of rehabilitation therapy and age

WeeFIM-C	Source	df	SS	MS	*F*	*P*	*Post hoc* tests[a]	η^{2b}
Self-care	Between groups	2	2518.94	1259.47				
	Within groups	68	15,815.3	232.58	5.42**	0.007	RT1 >RT0* RT2 >RT0**	0.14
	Total	70	18,334.2	1492.05				
Mobility	Between groups	2	1643.11	821.56				
	Within groups	68	6835.28	100.52	8.17**	0.001	RT1 >RT0** RT2 >RT0**	0.19
	Total	70	8478.39	922.08				
Cognition	Between groups	2	655.46	327.73				
	Within groups	68	6731.8	98.99	3.31*	0.042	-	0.09
	Total	70	7387.26	426.72				
Total score	Between groups	2	12,695.6	6347.79				
	Within groups	68	67,025.6	958.67	6.44**	0.003	RT1 >RT0* RT2 >RT0**	0.16
	Total	70	79,721.2	7306.46				

*, **Denote significance at the 5% and 1% levels, respectively, [a]*Post hoc* comparison with the Bonferroni method, η^{2b}: Small effect size=0.01; moderate effect size=0.06; large effect size=0.14. SS: Sums of squares, MS: Mean squares

and compared the effects of RT among the three groups (RT0, RT1, and RT2). We also demonstrated considerably differences in functional independence among three studied groups. These findings may suggest that RT should be provided at least once per week and begin shortly after a child is diagnosed with rare and/or genetic diseases, and psychomotor retardation without a confirmatory diagnosis.

The low involvement in RT for patients who receive it no more than once per month may be due to inconvenient transportation, intergenerational parenting, living habits, cultural issues, and low socioeconomic status in Eastern Taiwan [7,8]. Previous research has discovered that in Eastern Taiwan, rural residents spend 2.5 times as much time on transportation to get to medical institutions compared with nonrural residents. Unfavorable living conditions and issues associated with intergenerational parenting leads to a vicious cycle of poverty, illness, and care issues. Because scheduling outpatient RT activities requires resources and time management, it is recommended to inform patients who only receive RT no more than once a month about the value of home rehabilitation programs (e.g., improved access to equipment at home and home modifications) and to discuss it with their families.

Overall, our RT program produced excellent results, especially in the mobility and self-care domains; the program is proven to slow down or halt functional decline [2,5,6].

Study limitations

Although the RT1 and RT2 children typically received outpatient RT once or twice a week, effects from the intensity, duration, and frequency of the RT on functional performance were not observed. The finding could be explained by the small sample size and heterogeneous groups. Further research is needed to clarify whether RT, particularly once per week, can yield favorable outcomes for children with rare or genetic diseases.

The methodological limitations of our study should be considered when interpreting the current findings. Specifically, this study was a retrospective review of children from a single rehabilitation center. In this context, specific therapy delivery may be part of the center-specific procedures and policies, and this may have influenced functional outcomes. However, considering the low prevalence of rare and genetic diseases among children, a randomized, controlled trial with multiple sites and treatment models may overcome this limitation. Furthermore, we did not assess baseline data by WeeFIM-C. The use of the WeeFIM-C in clinical settings should be further studied for a more thorough comparison.

Conclusions

The results of this study demonstrated that the functional status was different among three studied groups in terms of self-care, mobility, and cognitive functioning after receiving outpatient RT. Therefore, such rehabilitation should begin shortly after diagnosis. Moreover, other alternatives such as home rehabilitation programs, greater access to equipment at home, and home modifications can be suggested to the families of such children who only receive RT no more than once a month. In addition, a randomized, controlled trial with multiple sites should be conducted to corroborate these findings. It is also essential to include the WeeFIM-C in clinical assessments to evaluate functional outcomes during outpatient RT.

Data availability statement

All data generated or analyzed during this study are included in this published article.

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Conflicts of interest

Dr. Shao-Yin Chu, an editorial board members at *Tzu Chi Medical Journal*, had no role in the peer review process of or decision to publish this article. The other authors declared no conflicts of interest in writing this paper.

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