# A case study of a patient with CMT4F based on ICF-CY framework

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# Abstract

This case is the first time to combine ICF-CY conceptual framework with GMFM-88 item functional scale and use it in the rehabilitation and diagnosis of early childhood onset patients with CMT4F.Through periodical training, the effectiveness to guide rehabilitation diagnosis and treatment for early onset CMT4F patients was proved.

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Abstract

BACKGROUND & OBJECTIVE: To explore the effectiveness of using ICF-CY framework and GMFM-88 functional assessment to analyze rehabilitation diagnosis and treatment of patients with CMT4F.

Case Description: The child is 3 years old and 11 months old, and was diagnosed with CMT4F type at 15 months old. In this case, ICF-CY framework and GMFM-88 were used to demonstrate the ideas of rehabilitation diagnosis and treatment and to explore the effectiveness of treatment.

Outcome; After half a year of systematic community rehabilitation training and family training, the children's short-term goals STG1, STG2 and long-term goals LTG have been completed, and all items of GMFM-88 score full marks.

Discussion: This case is the first time to combine ICF-CY conceptual framework with GMFM-88 item functional scale and use it in the rehabilitation and diagnosis of early childhood onset patients with CMT4F.Through periodical training, the effectiveness of ICF-CY framework combined with GMFM-88 evaluation to guide rehabilitation diagnosis and treatment for early onset CMT4F patients was proved.

Keywords: ICF-CY, GMFM-88, CMT4F, Rehabilitation

# 1 Introduction

Charcot-Marie-Tone (CMT), also known as hereditary motor and sensory neuropathy (HMSN), is the most common hereditary disease of the peripheral nerve, with an incidence of about 1/2500 (Reilly, 1998).The main clinical manifestations are progressive symmetrical distal weakness and muscular atrophy, hypoesthesia and weakened or absent tendon reflex (Skre, 1974).Most of the patients start in childhood and adolescence, and the first symptoms are mostly in the lower extremities, gradually progressing to the upper extremities. CMT is divided into subtypes based on neuroelectrophysiological and genetic patterns. Among all subtypes, the most common subtype was CMT1A, accounting for about 70% of the total number of patients with CMT (Nelis, 1999).The second is CMT2. CMT4 is the rarest and has clinical characteristics. For example, most patients start at a young age and progress rapidly. Due to different degrees of sensory disorders and spinal deformity, they have obvious distal limb deformity, and in severe cases they will eventually lose the ability to walk. According to the study, the clinical manifestations of patients diagnosed with CMT4F should be more severe CMT1, and these patients have significant sensory loss. In this study, a child diagnosed with CMT4F type was selected with no sensory impairment, mainly manifested as motor dysfunction (Casasnovas, Cano, Alberti, Cespedes, & Rigo, 2008).

ICF-CY is a globally recognized functional evaluation system. It makes a detailed analysis and comparison of patients from several aspects of body structure and function, activities, participation, personal and environmental factors, and provides a unified standard for relevant personnel in rehabilitation treatment to discuss patients' functional level.ICF-CY is commonly used as a rehabilitation treatment framework for common diseases, which involves the complete ICF core entry.In order to reflect the patient's function in more detail, ICF Qualifier is used as the scoring system.The purpose of this case is to use ICF-CY framework and GMFM-88 functional assessment to share the ideas of rehabilitation diagnosis and treatment of CMT4F type and explore its effectiveness.

#### 2. The Case Description

#### 2.1 Patient History

The child, male, aged 3 years and 9 months, was born on March 18, 2016. He was delivered at full term and naturally, the second child in his family. The first child was a girl with no abnormality. The mother recalled the pregnancy and the period of delivery without any special condition, and denied that the labor was too long, ischemia and hypoxia, and the fetal position was not right. Because "the child can only raise his head independently for 4+ months, can only sit alone for 7 months, still can not walk for 15 months" to the outpatient department of our hospital. At the time of routine child care in the local area, the child was diagnosed as "gross motor development lag?" For further screening, electromyography, cranial imaging and

genetic testing were performed, and the diagnosis was CMT4F type.Referral to rehabilitation department for routine rehabilitation assessment/treatment.

The patient was first evaluated in our hospital on June 22, 2017, and the evaluation indicated that the child's developmental level was slightly behind the normal range (the child could stand alone for a few seconds and dash forward a few steps). The children and their families participated in home rehabilitation training once, and the following training was mainly conducted in the local rehabilitation center (mainly PT training). At the fourth assessment on February 3, 2019, it was suggested that the child was slightly behind the normal range of development (poor balance coordination, poor right lower limb strength). The children were followed up for rehabilitation every year, during which rehabilitation assessment and family training were conducted. This case is the result of evaluation, treatment and reevaluation on August 1, 2020, after the rehabilitation every under the family training in our center on February 1, 2020, and the 6-month training in the local rehabilitation center. Through two evaluation results, the rehabilitation treatment effect of this disease under the framework of ICF-CY was discussed.

2.2 Assessment (see Table 1)

The main problems listed by the therapist in terms of body structure and function in ICF-CY included 1) B7301 "Power of One Limb" : the right leg supported for 3 seconds in GMFM-88 test (see Attached Table 2);2) B770 "Gait pattern functions" : By observing walking, the patient showed the disappearance of swinging arm, increased slosh, and wide base walking;3) B7409 "Muscle Endurance Functions, Unspecified" : In the endurance test, the child is prone to fatigue and cannot walk continuously for 6 minutes;4) B7602 "Coordination of voluntary movement" : Through GMFM-88 and observation of activities such as kickball and rope skipping, the Coordination of children was poor;5) S7709 "Additional Musculoskeletal structures related to movement, unspecified" : observation of the overall parataxel alignment shows that the parataxel alignment of the right lower limb in the child is poor when moving.

In terms of the activity and participation levels in ICF-CY, the main problems listed by the therapist included 1) D 4502 "Walking on different surfaces" : the child could not walk freely on rough or soft surfaces, and the balance ability during Walking was limited;2) D 4552 "RUNNING" : Children should not run at a variable speed or under dual tasks;3) D 4553 "Jumping" : the child jumps with both legs 10cm, and jumps with one leg on the right side 5cm.

Among the environmental and personal factors of ICF-CY, E310 "Immediate Family" means that the Family members of the child have a positive attitude towards the treatment, which is conducive to the treatment. E340 "Personal Care Providers and Personal Assistants" : accompanied by the child's family members (grandma);In terms of personal factors, the children themselves have strong subjective initiative and a more positive attitude towards rehabilitation. On the other hand, children with language communication barriers, extroverted personality, like to communicate with different people, is conducive to the establishment of children's confidence in the process of treatment.

All the relevant therapists in the rehabilitation team explained the overall situation of the patient. OT reported B140 "attention functions" and B152 "Emotional functions" because of the child's lack of attention and poor Emotional control ability. According to psychotherapists, D240 "Handling stress and other psychological demands" is difficult for the patient to deal with stress and other emotions. The family reported that D571 "Looking after one's safety" was unable to take care of their own safety when completing certain activities, and there were certain safety problems.

#### 2.3 Intervention

The rehabilitation goal of this case is established based on the ICF-CY framework and through the consultation of relevant responsible therapists, children, family members and caregivers (grandma). The children had 2 short-term goals, including 1) right leg jumping 10cm;2) Improve mobility in different environments (STG2). The long-term rehabilitation goal is that play is not affected, which takes into account the patient's age and growth characteristics. Based on short-term rehabilitation goal 1 (STG1) and ICF-CY framework analysis, the main training programs should focus on the physical structure and function of children, such as B455 Exercise tolerance functions, B730 Muscle power functions, B740 Muscle endurance functions, and B760 Control of voluntary movement functions. Programs include strength training, spa treatments, and therapeutic games. The short-term rehabilitation goal 2 (STG2) training mainly consisted of walking training, jumping training and running training. The individual and environmental factors such as the high degree of cooperation of the children's family members, the strong subjective initiative of the children, the cooperation of the family members and caregivers can promote the successful completion of rehabilitation training.

Rehabilitation professionals involved in rehabilitation training include PT, OT, sports therapists, community rehabilitation physicians, etc. As a child patient, family members and regular caregivers (grandma) are also the main participants. Carter GT team believed that patients with CMT needed the intervention of a multidisciplinary team, including neurologists, psychiatrists, orthopedic surgeons, physical therapists, occupational therapists, and orthopedic specialists (AgnesJani-Acsadi, 2008). The objective of this comprehensive team should be to maximize patient independence and improve the quality of life of patients (Carter, 2008). However, community rehabilitation centers have their own limitations, so the health professionals involved in this case still have limitations.

According to the particularity of individual cases, Intervention training will be divided into two parts: community rehabilitation training (see Table 3) and family rehabilitation training. According to the existing literature, the training intensity was selected as light to medium intensity (Chetlin, 2004; El Mhandi, 2007; Lindeman, 1995; Young P, 2008).

# 2.3.1 Intervention under ICF-CY Body Structure & Function

According to the results of the Assessment, in terms of the body structure and function of the ICF-CY framework, the main training programs and exercise doses are 1) strength training of the right lower limb in the community, including isokytic muscle strength training and elastic band muscle strength training, 4 times a week, with a duration of 30 minutes; Family strength training is mainly carried out on the end of the patient's limbs with extra weights of about 2kg, 3 times a week, and the training duration is 20-30 minutes.2) Walk training including treadmill walking training, walking part practice, 5 times a week, one time for 30 minutes;3) Spa, once or twice a week, for 30-45 minutes; 4) Therapeutic games, including jigsaw puzzles, toy placement and hide-and-seek, are inserted into each treatment process to improve the participation and interest of patients during the rest.

# 2.3.2 Intervention under ICF-CY Activity & Participation

According to the results of Assessment, at the activity and participation level of ICF-CY framework, the main training program and exercise dose are: 1) adaptive training on walking, 5 times a week, for 30 minutes, outside the treatment room, such as grass, slope and steps;2) Running training: on the 5-meter trail, running back and forth, variable speed running, weight bearing running, etc., 5 times a week, the training duration is 30 minutes;3) Jumping training, including hoop jumping, long jump, jumping obstacles, etc., 5 times a week, the training duration is 30 minutes.

#### 2.4 Outcome

After six months of systematic community rehabilitation training and family training, the child's function has been significantly improved. The evaluation results of follow-up were shown in Table 1. According to the body structure and function of ICF-CY frame, B7301 "Power of One Limb", the right leg support of the child could last from 3 seconds to 12 seconds, which changed from 2 points to full score in GMFM-88.B770 "Gait Pattern Functions", the walking posture of children is more close to nature, in line with the walking characteristics of this age group.S7709 "Additional Musculoskeletal structures related to movement, unspecified", children with lower limb running, jumping and other line lifting.

Activity and participation levels under the ICF-CY framework; D 4502 "Walking on different surfaces", the child can walk on any surface, especially uneven surfaces or grass surfaces. D 4552 "RUNNING", the child

can complete short distance variable speed Running. D 4553 "Jumping" means that the children's Jumping ability is improved, which can assist in Jumping trampolines or free Jumping on the ground. For long-term goals (LTG), children can play in kindergartens or playgrounds at this stage without any restrictions as long as it is safe.

#### Discussion

This case has individualized short - and long-term goals under the framework of ICF-CY.Through phased training (including strength training and functional training, etc.), the strength of the patient's unilateral limbs has been improved, which is consistent with the results reported in previous literature (Cup, 2007). At the same time, the improvement of movement ability of patients is mainly manifested in three aspects: walking posture, walking stability and walking endurance, namely, the improvement of alignment and alignment in children's movement, and the improvement of movement ability and aerobic metabolism ability in different environments (DavidD.Kilmer, 2002; Kilmer DD, 2005). It has been documented that patients wearing orthopedic insoles can improve foot alignment, increase foot support surface, increase the stability of foot structure and walking stability, ensure forward propulsion, synchronize movement and assist walking (Casasnovas et al., 2008), thus improving the quality of life of children with CMT.The child in this report began to use orthopedic insoles at the age of 2 and was reexamined every six months to ensure that the orthopedic insoles and shoes could support the child's walking and higher level of functional activities.

The ICF-CY framework was first proposed in 2007, and its main purpose is to document the limited participation of children. At the same time, it assists with the cooperation of health-professionals to assist children to maximize their participation in daily life activities (Granlund, 2013). However, in practice, because of the complexity of coding system, it has not been widely used. At present, ICF-CY framework is used to describe the rehabilitation of Spinal Muscular Atrophy (SMA), children with cancer, Cerebral Palsy (CP), adolescent vegetative state and children with visual impairment (Darcy L, 2015; Franki I, 2012; Leonardi M, 2012; Rainey L, 2014; Trabacca A, 2020).In this paper, CMT- Cy framework was first applied to children with CMT onset, and this classification is relatively rare in clinical practice.CMT patients with early childhood onset also need to consider infant development, such as the presence of milestones of gross motor function.Therefore, ICF-CY alone could not accurately reflect the overall condition of the child, so this case adopted the ICF-CY framework combined with GMFM-88 items for case analysis.Burak and Kavlak demonstrated that quality of life, activities and participation, and environmental factors were correlated under the ICF-CY framework in adolescent children with cerebral palsy.However, the subjects selected in this paper are adolescent patients, and there are many differences between them and infant patients still in the growth and development stage. Therefore, GMFM-88 combined with ICF-CY framework is a highlight of this paper.

There is no unified treatment strategy and prognosis judgment in all literatures (Pareyson D, 2009). At present, there is no effective medical treatment, supportive care is the most common management mode and is limited to rehabilitation and orthopedic surgery for musculoskeletal problems. Rehabilitation requires teamwork, especially between neurologists and physiotherapists. Existing literature shows that 70% of patients with CMT have sensory dysfunction, such as decreased vibration and joint position, decreased pain or temperature, and garty-like sensory distribution disorder. This case did not have similar sensory problems, and the study on CMT4F classification of early onset was blank.

In addition, because of the slow progression of symptoms in patients with CMT, long-term follow-up of these patients is particularly important. There are a few literatures on the effectiveness of long-term rehabilitation of CMT (Sman AD, 2015). Due to the influence of many factors such as experimental quality and sample size, high-quality studies are needed to confirm its importance. Article proves that with the progress of the disease, patients with juvenile onset of CMT ankle deformity may occur and the surrounding muscle contracture, and so on and so forth, and the patients in the case of juvenile onset, the degree of disease progression is unknown, therefore, the meaning of the long-term follow-up is not only with the growth of musculoskeletal problems also need to know for the unique performance of the disease, the effectiveness of rehabilitation method.

Although the effect of rehabilitation therapy has been confirmed in some literatures, there is no unified clinical rehabilitation guideline and exercise prescription for CMT disease (Cup, 2007), including training amount, training frequency, training duration and training method (Sman AD, 2015). Therefore, the training prescription in this case was mainly derived from the conventional treatment dose.

According to the Cieza team, the importance of rehabilitation therapy is still underappreciated globally, and in some poor parts of the world, rehabilitation is still considered as a form of post-operative treatment. According to statistics, one out of every three people in the world (about 2.45 billion people) need rehabilitation treatment when suffering from illness or injury (Cieza A, 2021). However, most countries and regions cannot meet the needs of rehabilitation, and rehabilitation resources are still very scarce. For the purpose of rehabilitation therapy, people's cognition is still at the level of recovery function, and rehabilitation therapy can not only optimize individual function to the greatest extent, but also improve the ability of education, employment, home independence and other abilities of patients.

As for treatment strategies, the rehabilitation treatment of common diseases is guided by clinical rehabilitation guidelines, and the effectiveness of rehabilitation has been demonstrated by a large number of studies. However, for rare diseases, such as CMT4F, there is no recognized clinical rehabilitation guideline, and only a few literatures have confirmed the effectiveness of strength training, balance training, endurance training and functional training for this disease (Lindeman E, 1999; Lindeman, 1995; Ramdharry GM, 2014). The need for rehabilitation may be long-term, or even lifelong, and requires the consumption of a large number of human, material and financial resources of the family, society and the country. The Cieza team also proposed that rehabilitation should gradually be more community-based and benefit more patients (Cieza A, 2021).Therefore, the routine rehabilitation treatment of this case was carried out in the community rehabilitation center, which made the distribution of rehabilitation more reasonable.

#### Limitation

There are limitations to this report. First of all, this case only reported the children's community and family sports training program, and did not describe the sensory integration training, hydrotherapy and other training in detail, and lacked a detailed description of multidisciplinary comprehensive rehabilitation. Secondly, this case only reported a single evaluation and a follow-up after half a year, and no long-term follow-up was conducted. In view of the characteristics of slow disease progression, long-term rehabilitation training and follow-up should be carried out to better evaluate the effectiveness of rehabilitation for this type of patients; Finally, CMT4F, as a rare clinical case, puts great mental and economic pressure on the children and their families when they are diagnosed. The families of the children have a very limited understanding of the disease. In order to reduce the pressure of the children and their families, and make the family members and the treatment team better cooperate, genetic counseling services should be provided. Studies have proved that appropriate medical information and genetic counseling are crucial for children with CMT and their families (ME., 2006; Szigeti K, 2009).

# Conclusion and clinical implications

In this case, ICF-CY conceptual framework and GMFM-88 item functional scale were combined for the first time to be applied in the rehabilitation process of early childhood onset CMT4F patients. Through periodical training, the effectiveness of ICF-CY framework combined with GMFM-88 evaluation to guide rehabilitation diagnosis and treatment for early onset CMT4F patients was proved.

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# CONFLICT OF INTEREST

The authors declare that they have no conflict of interest.

# AUTHOR CONTRIBUTIONS

Jing Zhang drafted the manuscript. Longyuan He collected data and restored data. Zhihui Li collected data and underwent data analysis. Lv Wei analyzed all data thoroughly. Jing He supervised the writing of the manuscript. All authors read and approved the final manuscript.

# ETHICAL APPROVAL

Ethical approval was obtained from the biomechanical ethics committee of West China Hospital at Sichuan University.

# WRITTEN CONSENT FROM THE PATIENT

Written informed consent was obtained from the patient's Next of kin for publication of this case report.

#### DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available from the corresponding author upon reasonable request.

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Table 1. Physical Therapist Documentation Template: Selected Codes as Examples

Patient rehabilita- tion goal: play is not affected Short- term Goals (STG) : Jump 10cm on one leg on the right side; 2) The ability to move in different environ- ments Long-term goal (LTG) : Transnis- tria of leisure activities	Patient rehabilita- tion goal: play is not affected Short- term Goals (STG) : Jump 10cm on one leg on the right side; 2) The ability to move in different environ- ments Long-term goal (LTG) : Transnis- tria of leisure activities	Patient rehabilita- tion goal: play is not affected Short- term Goals (STG) : Jump 10cm on one leg on the right side; 2) The ability to move in different environ- ments Long-term goal (LTG) : Transnis- tria of leisure activities	Patient rehabilita- tion goal: play is not affected Short- term Goals (STG) : Jump 10cm on one leg on the right side; 2) The ability to move in different environ- ments Long-term goal (LTG) : Transnis- tria of leisure activities	Diagnosis: CMT4F type	Diagnosis: CMT4F type	Diagnosis: CMT4F type	Diagnosis: CMT4F type		
The date of	February 1, 2020 Examination	February 1, 2020 Examination	February 1, 2020 Examination	February 1, 2020 Examination	February 1, 2020 Examination	February 1, 2020 Examination	February 1, 2020 Intervention + Frequency	August 1, 2020 Re- examination	Au 1, Re ex
ICF Categories Interven- tion Target	The Need to Examine?	Test	Test Value	Test Value	Intervention Target, Related to STG/LTG Number?	Prognosis (Only for Interven- tion Targets)	Physical Therapy Sessions Will Be Provided 1 or 2 Times Daily	Retest Value (Only for Interven- tion Targets)	Go Ao
Body Functions b7301 Power of mus- cles of one limb	Body Functions Yes	Body Functions GMFM- 88	Body Functions Hold on the right foot for 3 seconds	Body Functions Hold on the right foot for 3 seconds	Body Functions STG1	Body Functions Strength train- ing might in- crease muscle power	Body Functions Strength train- ing daily in the community	Body Functions Hold on the right leg for 12 seconds	Bo Fu Ye

b770 Gait Pattern Functions	Yes	Observationa Gait Analysis	alLack of arm swing Unsteady walking Large base of support	Lack of arm swing Unsteady walking Large base of support	STG1	Walking improvement	Gait training indoors and outdoors	With arm swing Reason- able base of support	Ye
b7409 Muscle en- durance func- tions, unspecified	Yes	Endurance test	Six min- utes walk test	Six min- utes walk test	STG1/STG2	Muscle en- durance increased	Water walk- ing, en- durance walking	Walking 2km and climb- ing 5 stairs has no muscle pain	Ye
b7602 Coor- dina- tion of volun- tary movement	Yes	Observation- 88	+Kikkfild- ball, skip- ping rope and other activi- ties are not accordinated	Kicking ball, skip- ping rope and other activi- ties are not accordinated	STG1	Coordination improved	Coordination training	activi- ties such as playing foot- ball with high quality	Ye
Body Structure s7709 Addi- tional muscu- loskele- tal struc- tures related to move- ment, unspecified	Body Structure Yes	Body Structure observation	Body Structure Alignment of lower extremities	Body Structure Alignment of lower extremities	Body Structure STG1	Body Structure Good alignment	Body Structure Part practice	Body Structure Lower ex- tremi- ties run- ning, jump- ing and other line lifting	Bo Str Ye
Activities and Participation	Activities and Participatio	Activities and n Participation	Activities and Participation	Activities and Participation	Activities and Participation	Activities and Participation	Activities and Participation	Activities and Participation	Ac an Pa

d 4502 Walk- ing on differ- ent surfaces	Yes	Observation	Do not walk freely on rough or soft ground	Do not walk freely on rough or soft ground	STG1	Balance improvement	Up and down the hill, walk- ing on the sponge	Can walk on any road surface	Ye
d 4552 Running	Yes	GMFM- 88	Do not run at a speed change or with dual tasks	Do not run at a speed change or with dual tasks	STG2	Improving Run- ning Ability	Variable speed run- ning, walk- ing and run- ning com- bined training	Variable speed run- ning can be com- pleted under supervision	Ye
d 4553 Jumping	Yes	GMFM- 88	Jump 10cm on both legs and 5cm on one leg on the right side	Jump 10cm on both legs and 5cm on one leg on the right side	STG1	Jumping ability improved	Assist jump- ing trampoline	Free jumping	Ye
Environment Factors	aInfluence	Influence	Sido	Side					
e310 Imme- diate Family e340 Per- sonal care providers and per- sonal	Positive Square root Square root	Negative							
assistants Personal Factors	Influence	Influence							
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Application of the ICF in Multidisciplinary Patient Management

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 Table 3 ICF-CY Intervention Table

Body	Intervention	n Interventio	n Intervent	ion Interventio	on				
Struc-	Targets-	Targets-							
ture &	ICF	ICF							
Functions	Categories	Categories							
	-	-	Type	Intensity	MD	$\mathbf{PT}$	ОТ	Others	Evaluation Value

	b7301	Power of muscles of one limb	Strength training with equip- ment Strength training with weights (HED)	4 / wk, a 30 mins 3/wk, 20~30mins	[?]			2
	b770	Cait	(IIEI ) Cait	РТ	[?]		[?]	3
	0110	Pat-	training	nro-	[•]		[.]	5
		tern	training	gram.				
		Functions		5/wk.				
				30mins				
	B455	Exercise	Aquatic	1-			[?]	2
		toler-	training	2/wk,				
		ance	-	30-				
		functions		45mins				
	b7602	Coordinati	onTherapeut	m ic5/wk,		[?]		1
		of vol-	games	30mins				
		untary						
	1 (500	movement	<b>a</b> .	DT	[0]			2
Activity	d 4502	Walking	Gait	PT	[?]			2
& D (*** )		on dif-	training	pro-				
Participat	ion	ferent		gram:				
		surfaces		O/WK,				
	d 4559	Bunning	Bunning	DT	[2]			2
	u 4002	Running	training	I I	[•]			0
			training	gram.				
				5/wk				
				30mins				
	d 4553	Jumping	Jumping	PT	[?]			2
		1 0	training	pro-				
			0	- gram:				
				5/wk,				
				30mins				

Schedule 2. The fifth assessment was completed on 1 February 2020.

# GMFM - 88

Functional partition	score	Total score	The percentage
A. Decubitus position and roll over	51	51	100%
B. seat	60	60	100%
C.Climb and jump	42	42	100%
D. standing	38	39	97%
E. Walk, run, jump	70	72	97%

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According to this scale, the child can go up and down stairs alternately alone, with poor stability; It can run and jump, but the knee joint and ankle joint are poorly controlled in the process of running. It needs to complete the one-legged jumping with assistance, and the posture control is poor during the jumping process, and the stability decreases obviously.