Measurement of the Functional Status of Patients with Different Types of Muscular Dystrophy

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Muscular dystrophy (MD) comprises a group of diseases characterized by progressive muscle weakness that induces functional deterioration. Clinical management requires the use of a well-designed scale to measure patients' functional status. This study aimed to investigate the quality of the functional scales used to assess patients with different types of MD. The Brooke scale and the Vignos scale were used to grade arm and leg function, respectively. The Barthel Index was used to evaluate the function of daily living activity. We performed tests to assess the acceptability of these scales. The characteristics of the different types of MD are discussed. This was a multicenter study and included patients diagnosed with Duchenne muscular dystrophy (DMD) (classified as severely progressive MD), Becker muscular dystrophy (BMD), limb girdle muscular dystrophy (LGMD) and facioscapulohumeral muscular dystrophy (FSHD). BMD, LGMD, and FSHD were classified as slowly progressive MD. The results demonstrated that the Brooke scale was acceptable for grading arm function in DMD, but was unable to discriminate between differing levels of severity in slowly progressive MD. The floor effect was large for all types of slowly progressive MD (range, 20.0–61.9), and was especially high for BMD. The floor effect was also large for BMD (23.8%) and FSHD (50.0%) using the Vignos scale. Grades 6–8 of the Vignos scale were inapplicable because they included items involving the use of long leg braces for walking or standing, and some patients did not use long leg braces. In the Barthel Index, a ceiling effect was prominent for slowly progressive MD (58.9%), while a floor effect existed for DMD (17.9%). Among the slowly progressive MDs, FSHD patients had the best level of functioning; they had better leg function and their daily living activities were less affected than patients with other forms of slowly progressive MD. The results of this study demonstrate the acceptability of the different applications used for measuring functional status in patients with different types of MD. Some of the limitations of these measures as applied to MD should be carefully considered, especially in patients with slowly progressive MD. We suggest that these applications be used in combination with other measures, or that a complicated instrument capable of evaluating the various levels of functional status be used.

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Muscular dystrophy (MD) comprises a group of genetic diseases that is marked by progressive muscle weakness and atrophy. Various gene deficits cause different clinical manifestations. Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD) are both caused by defects in the Xp21 region of the X chromosome [1]. The incidence of DMD is about 1 in 3,500 male births; the symptoms present during childhood and lead to rapid deterioration [2]. Other types of MD are less common, such as facioscapulohumeral muscular dystrophy (FSHD) and limb girdle muscular dystrophy (LGMD). The flawed gene for FSHD is located on chromosome 5q35 and shows autosomal dominant inheritance. The condition is characterized by weakness of the face, upper-arm and shoulder girdle muscles [3]. LGMDs comprise a group of genetic disorders mainly involving the pelvic and/or shoulder girdle musculatures, and where transmission is due to both autosomal recessive and dominant patterns [4–6]. Many clinicians and therapists assess function as a means of determining changes [7,8]. The suitability of the scales used for evaluating the functional status is therefore an important issue in patients with MD.

The Brooke and Vignos scales provide ordinal-level data to assess the upper and lower extremity functions, respectively [9,10]. These scales were initially designed for DMD and many studies have used them to evaluate functional status [11–13]. They are now also used to evaluate the functional status of patients with neuromuscular diseases [14–16].

Use of the Brooke and Vignos scales to measure function in other types of MD is still limited and, although previous studies have included patients with various types of MD, a detailed analysis of the acceptability of these scales for different types of MD is still lacking. Acceptability is determined by examining whether or not the score distributions adequately represent the true distribution of functional status in the sample [17,18]. Because different types of MD were included within the same group in previous studies, it was not possible to demonstrate the real functional profile of the specific types of MD.

In addition to examining upper and lower extremity functions, independent activities of daily living (ADL) of patients are also important. The Barthel Index (BI) is a simple, well-established reliable and valid scale, and is one of the most widely used measurement tools in rehabilitation centers. BI scores are also used by the Bureau of Employment and Vocational Training in Taiwan as criteria when hiring foreign laborers as caregivers. Previous studies have used the BI to assess the performance of ADL in DMD [19–21]. However, its acceptability for use in different types of MD has never been studied.

Assessing the acceptability of these scales for evaluating different types of MD could allow for precise application of the scales. The aim of this study was therefore to assess the acceptability of the scales for evaluating the functional status of patients with various types of MD.

Patients and Methods

Subjects

Patients with MD with a range of severities were recruited from outpatient and inpatient units of the neurological departments of hospitals participating in a multicenter registration program for neuromuscular disease and in the development of an assessment tool program for muscular dystrophy in Taiwan. Four types of MD were included in this study: DMD, BMD, FSHD, and LGMD. A definite diagnosis of MD was confirmed by two neurologists on the basis of clinical and genetic analyses, and electromyographic and muscle biopsy criteria, according to the diagnostic criteria for neuromuscular disorders from the European Neuromuscular Research Group used at that time [14,15,21–24]. Patients were excluded if they had any other coexisting medical or psychiatric diseases. After diagnosis, patients were followed-up at outpatient clinics for at least 2 years to confirm the diagnosis. The study was approved by the hospital's institutional review board, and written informed consent was obtained from all participants.

Procedure

After diagnosis and follow-up, patients were evaluated for functional status by four trained raters (2 physical therapists and 2 occupational therapists). Most of the patients were evaluated in hospital, and those who had difficulties in getting to the hospital were evaluated by home visit. The Brooke scale was applied for grading upper extremity function [9] and the Vignos scale was used for lower extremity function [10]. The BI was used to evaluate ADL ability [25].

Instruments

Brooke and Vignos scales

The grades on the Brooke scale range from 1 to 6; 1 means that the patient is able to start with arms at the sides and can abduct the arms in a full circle until they touch above the head, while 6 means that they are unable to raise their hands to their mouth and have no useful function of the hands. On the Vignos scale, the grade ranges from 1 to 10; 1 means that the patient is able to walk and climb stairs without assistance, while 10 means that the patient is bed-bound (Table 1). Both the Brooke and Vignos scales are reliable with high intraclass correlation values for DMD [26].

BI

The BI is a 10-item scale that measures functional independence in the domains of personal care and mobility. For each item, patients receive a score of 0 if they are unable to carry out the activity, a maximum score if able to perform the activity independently, and an intermediate score if able to perform the activity with assistance. Possible values are assigned in increments of 5. The totals can range from 0 to 100. The reliability and validity of the BI administered by oral interview, observation or telephone interview has been well-established [27,28]. The total scores are broken down into the following severity categories: 0–20 represents totally dependent, 21–61 severely dependent, 62–90 moderately dependent, 91–99 mildly dependent, and 100 represents totally independent [29].

Data management and statistical analyses

To assess the acceptability of the instruments, missing data, observed versus possible score ranges, floor and ceiling effects, and skewedness were analyzed. The floor and ceiling effects were the proportions of patients with the minimum and maximum scores, respectively. The skewedness measured the symmetry of the sample distribution [18,30].

Brooke scale, Vignos scale, and BI scores were compared between severely progressive MD (DMD) and slowly progressive MD (BMD, LGMD, FSHD) using the Mann-Whitney U test. Brooke scale, Vignos scale, and BI scores were compared among the slowly progressive MDs (BMD, LGMD, FSHD) using the nonparametric Kruskal-Wallis one-way analysis of variance. The significance level was set at 0.05. All statistical analyses were done with SPSS version 10.0 (SPSS Inc., Chicago, IL, USA) for Windows.

Results

The study population comprised 179 patients from Northern, Central, and Southern Taiwan. Eighty-four patients (46.9%) were diagnosed with DMD and

Table 1. Grading for Brooke and Vignos scales

Grade	Functional description
	Brook scale for upper extremities
1	Starting with arms at the sides, the patient can abduct the arms in a full circle until they touch above the head
2	Can raise arms above head only by flexing the elbow (shortening the circumference of the movement) or using accessory muscles
3	Cannot raise hands above head, but can raise an 8-oz glass of water to the mouth
4	Can raise hands to the mouth, but cannot raise an 8-oz glass of water to the mouth
5	Cannot raise hands to the mouth, but can use hands to hold a pen or pick up pennies from the table
6	Cannot raise hands to the mouth and has no useful function of hands
	Vignos scale for lower extremities
1	Walks and climbs stairs without assistance
2	Walks and climbs stair with aid of railing
3	Walks and climbs stairs slowly with aid of railing (over 25 seconds for 8 standard steps)
4	Walks unassisted and rises from chair but cannot climb stairs
5	Walks unassisted but cannot rise from chair or climb stairs
6	Walks only with assistance or walks independently with long leg braces
7	Walks in long leg braces but requires assistance for balance
8	Stands in long leg braces but unable to walk even with assistance
9	Is in a wheelchair
10	Is confined to a bed

were classified as having severely progressive MD. Ninety-five patients (54.1%) were classified as having slowly progressive MD, and included 21, 54 and 20 patients, respectively, with BMD, LGMD, and FSHD. The demographic data for these patients with different types of MD are shown in Table 2. All patients with DMD or BMD, 60% of those with FSHD, and 59.3% of those with LGMD were male. The mean age of patients with DMD was the lowest at about 12 years, while the mean age of patients with LGMD was the highest at about 39 years. The mean disease duration for the various types of MD ranged from 9.3 to 17.4 years.

The measures were easy to assess and it took about 5–10 minutes to complete the tests, and patients did not feel uncomfortable. Less than 5% missing data is considered to be acceptable [17], and no missing data were found for any measures in this study.

The grade distributions for the Brooke and Vignos scales are shown in Table 3. Most of the patients (n = 135, 75.4%) were graded 1–3 for upper extremity function using the Brooke scale, while using the Vignos

Table 2. Characteristics of patients with different types of muscular dystrophy

	Total sample	DMD	BMD	FSHD	LGMD
Number (male/female)	179 (149/30)	84 (84/0)	21 (21/0)	20 (12/8)	54 (32/22)
Age (yr)	24.3 ± 16.5 (6–80)	12.4 ± 4.6 (6–26)	22.9 ± 9.8 (9–44)	35.5 ± 16.7 (13–80)	39.3 ± 15.6 (12–76)
Disease duration (yr)	13.1 ± 10.1 (2–70)	9.3 ± 4.8 (2–25)	13.4 ± 9.2 (2–32)	17.3 ± 14.5 (4–70)	17.4 ± 12.2 (2–64)
Brooke scale (range)	2.7 ± 1.7 (1–6)	3.2 ± 1.9 (1–6)	1.9 ± 1.5 (1–6)	2.3 ± 0.8 (1–3)	2.3 ± 1.3 (1–5)
Vignos scale (range)	5.2 ± 3.4 (1–10)	7.1 ± 3.1 (1–10)	3.5 ± 2.7 (1–10)	2.2 ± 1.3 (1–5)	3.9 ± 2.7 (1–9)
Barthel Index (range)	67.7 ± 35.6 (0–100)	44.3 ± 34.4 (0–100)	86.0 ± 29.1 (5–100)	97.8 ± 4.7 (85–100)	85.6 ± 20.2 (5–100)

DMD = Duchenne muscular dystrophy; BMD = Becker muscular dystrophy; FSHD = facioscapulohumeral muscular dystrophy; LGMD = limb girdle muscular dystrophy.

Table 3. Distribution of Brooke and Vignos scales for different types of muscular dystrophy

	Total sample	DMD	BMD	FSHD	LGMD
Brooke					
1	65 (36.3%)	28 (33.3%)	13 (61.9%)	4 (20%)	20 (37%)
2	26 (14.5%)	6 (7.1%)	4 (19.0%)	6 (30%)	10 (18.5%)
3	44 (24.6%)	15 (17.9%)	1 (4.8%)	10 (50%)	18 (33.3%)
4	8 (4.4 %)	8 (9.5%)	0	0	0
5	18 (10.1%)	10 (11.9%)	2 (9.5%)	0	6 (11.1%)
6	18 (10.1%)	17 (20.2%)	1 (4.8%)	0	0
Vignos					
1	27 (15.1%)	4 (4.8%)	5 (23.8%)	10 (50%)	8 (14.8%)
2	26 (14.5%)	9 (10.7%)	5 (23.8%)	2 (10%)	10 (18.5%)
3	32 (17.9%)	8 (9.5%)	4 (19.0%)	4 (20%)	16 (29.6%)
4	13 (7.3%)	2 (2.4%)	2 (9.5%)	3 (15%)	6 (11.1%)
5	9 (5.0%)	3 (3.6%)	2 (9.5%)	1 (5%)	3 (5.6%)
6	4 (2.2%)	3 (3.6%)	0	0	1 (1.9%)
7	0	0	0	0	0
8	1 (0.6%)	1 (1.2%)	0	0	0
9	51 (28.5%)	39 (46.4%)	2 (9.5%)	0	10 (18.5%)
10	16 (8.9%)	15 (17.9%)	1 (4.8%)	0	0
Total number	179	84	21	20	54

DMD = Duchenne muscular dystrophy; BMD = Becker muscular dystrophy; FSHD = facioscapulohumeral muscular dystrophy; LGMD = limb girdle muscular dystrophy.

Table 4. Acceptability of Brooke and Vignos scales for different types of muscular dystrophy

	Mean ± SD	Observed score (range)	Skewedness	Floor effect (%)	Ceiling effect (%)
Brooke scale					
Total sample	2.7 ± 1.7	1–6	0.7	36.3	10.1
DMD	3.2 ± 1.9	1–6	0.2	33.3	20.2
Slowly progressive MD	2.2 ± 1.3*	1–6	1.0	38.9	1.1
BMD	1.9 ± 1.5	1–6	1.8	61.9	4.8
FSHD	2.3 ± 0.8	1–3	–0.6	20.0	0
LGMD	2.3 ± 1.3	1–5	–0.2	37	0
Vignos scale					
Total sample	5.2 ± 3.4	1–10	0.2	15.1	8.9
DMD	7.1 ± 3.1	1–10	–0.8	4.8	17.9
Slowly progressive MD	3.4 ± 2.6†	1–10	1.3	24.2	1.1
BMD	3.5 ± 2.7	1–10	1.4	23.8	4.8
FSHD	2.2 ± 1.3	1–5	0.7	50.0	0
LGMD	3.9 ± 2.7	1–9	1.1	14.8	0

*$p = 0.001$, †$p < 0.001$ (slowly progressive MD compared with DMD by Mann-Whitney U test). DMD = Duchenne muscular dystrophy; BMD = Becker muscular dystrophy; FSHD = facioscapulohumeral muscular dystrophy; LGMD = limb girdle muscular dystrophy.

scale for lower extremity function, 51 patients (28.5%) were grade 9 and confined to wheelchairs. Only five (2.8%) patients were graded 6–8 and they used long leg braces for walking or standing.

The acceptabilities of the Brooke and Vignos scales are shown in Table 4. The mean Brooke scores were 3.2 (DMD), 1.9 (BMD), 2.3 (FSHD) and 2.3 (LGMD), and the mean Vignos scores were 7.1 (DMD), 3.5 (BMD), 2.2 (FSHD) and 3.9 (LGMD). The floor effect in the Brooke scale was large for all types of MD, ranging from 20.0 to 61.9, and was especially high in BMD patients. The ceiling effect was slightly increased (20.2%) for patients with DMD. The floor effect in the Vignos scale was large for patients with BMD (23.8%) and FSHD (50.0%), while for patients with DMD, the ceiling effect was also slightly increased (17.9%). The acceptable skewedness criterion was –1 to +1 [30]. The skewedness of the Brooke scale for all types of MD was 0.7, ranging from –0.6 to 1.8 for each type of MD, and that of the Vignos scale for all types of MD was 0.2, ranging from –0.8 to 1.4 for each type of MD.

The acceptability of the BI and the indicated dependence severity are shown in Table 5. Most patients with DMD were totally ($n = 23$, 27.4%) or severely ($n = 35$, 41.7%) dependent, while most patients with BMD, FSHD, and LGMD were totally independent (59%). When applied to the summary scores, floor and ceiling effects > 15% were considered to be significant [31]. The ceiling effect was 34.1% based on all types of MD, and 58.9% for slowly progressive MD (range, 48.1–80.0%). The floor effect still existed for patients with DMD (17.9%) but the ceiling effect was only 6.0%. Skewedness for all types of MD was –0.7, ranging from –2.3 to 0.3 for each type of MD.

The mean scores on the Brooke scale (3.2 ± 1.9), Vignos scale (7.1 ± 3.1), and BI (44.5 ± 34.4) for patients with DMD (severely progressive MD) were significantly different from those with slowly progressive MD (Brooke scale, 2.2 ± 1.3; Vignos scale, 3.4 ± 2.6; BI, 88.2 ± 20.1) by the Mann-Whitney U test ($z = -3.2$, $p = 0.001$; $z = -6.8$, $p < 0.001$; and $z = -8.5$, $p < 0.001$, respectively; Tables 4 and 5). There were no significant differences in upper extremity function among the different types of slowly progressive MD ($\chi^2 = 4.8$, $p = 0.09$), but lower extremity function and ADL were significantly different ($\chi^2 = 7.4$, $p = 0.03$; and $\chi^2 = 7.5$, $p = 0.02$, respectively).

Discussion

This study included patients with various types of MD and evaluated their functional status using the Brooke and Vignos scales and the BI. These scales were quick and easy to apply as means of measuring the functional status of patients with MD, and no missing data were found. In patients with slowly progressive MD, leg function was seldom graded 6, 7 or 8 using the Vignos scale, and arm function was seldom

Table 5. Acceptability of Barthel Index and indicated severity of dependence for different types of muscular dystrophy

	Acceptability				Number of patients by dependence severity				
	Mean ± SD	Observed score (range)	Skewedness	Floor effect (%)	Ceiling effect (%)	0–20	21–61	62–90	91–99	100
Total sample	67.7 ± 35.6	0–100	–0.7	8.4	34.1	25	44	34	15	61
DMD	44.5 ± 34.4	0–100	0.3	17.9	6.0	23	35	11	10	5
Slowly progressive MD	88.2 ± 20.1*	5–100	–2.3	0	58.9	2	9	23	5	56
BMD	86.0 ± 29.1	5–100	–2.1	0	66.7	1	2	2	2	14
FSHD	97.8 ± 4.8	85–100	–1.8	0	80.0	0	0	4	0	16
LGMD	85.6 ± 20.2	5–100	–1.8	0	48.1	1	7	17	3	26

*$p < 0.001$ (slowly progressive MD compared with DMD by Mann-Whitney U test). DMD = Duchenne muscular dystrophy; BMD = Becker muscular dystrophy; FSHD = facioscapulohumeral muscular dystrophy; LGMD = limb girdle muscular dystrophy; 0–20 = totally dependent; 21–61 = severely dependent; 62–90 = moderately dependent; 91–99 = mildly dependent; 100 = totally independent.

graded 4, 5 or 6 using the Brooke scale. The BI showed a prominent ceiling effect in slowly progressive MD, and a floor effect in patients with DMD.

The Brooke scale was the best available measure for grading arm function in patients with severely progressive MD (DMD), but was not appropriate for those with slowly progressive MD (BMD, LGMD, FSHD). Most patients with slowly progressive MD were graded 1–3, which indicated that grades 4–6 were too simple for most of these cases. Integrated, more functional assessment items, may be needed to discriminate arm function, especially high level functional activity, in slowly progressive MD.

Vignos grades 6–8 were inapplicable in some cases because these grades covered the ability to walk with or without assistance and standing with a long leg brace. This could have been due to cultural differences in the use of long leg braces, but other studies also found that only a few patients were graded between 6 and 8, and they felt uncomfortable walking with a long leg brace [11,32]. When a patient is unable to walk independently and does not use a long leg brace for walking or standing, the leg function will change from grade 5 to grade 9. Although we strongly agree that using a long leg brace is an important treatment option for patients with MD, possible problems in assessing the leg function of grades 6–8 need careful consideration.

The acceptability of the BI differed between severely and slowly progressive MDs. A prominent ceiling effect was found in slowly progressive MD, whereas a floor effect was found in severely progressive MD. Ceiling effects for the BI have been found in many studies [33–36]. The BI may have limited ability to discriminate between outcomes in individuals with slowly progressive MD. Of the different types of slowly progressive MD, FSHD was associated with the best leg function and ADL was less affected than in the other types of MD.

A possible reason for the existence of the floor effect seen in DMD may be that those patients were very weak and lacked even basic mobility, such as the ability to roll or to move from a supine to a sitting position during the end stage of the disease. However, the BI is not designed for lower level mobility items. The use of some basic mobility items for patients with severely progressive MD and some higher level functional items for those with slowly progressive MD may therefore be needed.

A comprehensive scale able to evaluate the varied clinical problems is an important tool. The Muscular Dystrophy Functional Rating Scale was designed to fit the gap in assessing patients with various types of MD, and the scale has been demonstrated to have good reliability and validity [7,37].

Several limitations of our study are worth noting. One limitation was the lack of a reliability and validity study. Reliability and validity are important for determining the stability and confirming that the content of a scale represents the concept that it is intended to measure. The second limitation was that our sample only included patients with DMD, BMD, FSHD, and LGMD; other types of MD may present with different clinical profiles. The third limitation was that the subtypes of LGMD were not identified in our study. LGMDs are a heterogeneous group of inherited diseases and 19 different subtypes have been reported [38]. Subtypes of LGMD show clinical variability and difficulties in classification arise due to common sporadic cases [39,40]. Ethnic clusters and specific geographic origins are commonly found for certain subtypes of LGMD, such as LGMD 2A, which is the most prevalent form in Japan, Turkey, and Brazil [41–43]. Future studies in Taiwan are needed to investigate the types of genetic deficiencies and to explore the influence of functional performance in patients with muscular dystrophy.

This study investigated the Brooke and Vignos scales, and the BI as instruments for measuring the functional status of patients with different types of MD. This study included a large sample of patients with MD and demonstrates that the Brooke scale and BI are too simple to identify functional changes in patients with slowly progressive MD. For patients with DMD, the addition of basic mobility items may be needed to evaluate their function, due to the floor effect of the BI.

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不同類型肌肉失養症功能狀態之評估

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肌肉失養症 (**muscular dystrophy**) 是一群漸進性肌肉無力疾病，並造成功能持續退步。使用設計良好的評估表來正確評估功能狀態，為治療重要的基礎。本研究目的為檢測評估功能工具，使用於不同類型肌肉失養症功能狀態其合適情形。以 **Brooke** 和 **Vignos** 評估表分別評估上肢及下肢功能等級，以巴歇爾指數 (**Barthel index**) 評估日常活動的功能。統計檢測各種評估工具對不同類型肌肉失養症的接受度 (**acceptability**)，並討論不同類型的表現。病患來自台灣多個醫療院所，共有 **179** 位，**46.9%** 為裘馨型肌肉失養症 (**Duchenne muscular dystrophy**)，屬於嚴重進展的肌肉失養症；其餘 **53.1%** 為貝克型肌肉失養症 (**Becker muscular dystrophy**)、肢帶型肌肉失養症 (**limb girdle muscular dystrophy**) 與顏肩肱型肌肉失養症 (**fascioscapulohumeral muscular dystrophy**)，屬於緩慢進展的肌肉失養症。結果顯示 **Brooke** 評估表適用於裘馨型肌肉失養症，但不易分辨緩慢進展的肌肉失養症，其地板效應 (**floor effect**) 都較大 (範圍為 **20.0%** 至 **61.9%**)，尤其是貝克型肌肉失養症。**Vignos** 評估表方面，同樣的貝克型 (**23.8%**) 與顏肩肱型 (**50.0%**) 肌肉失養症有較大的地板效應，除此以外其評估等級 **6** 到 **8**，有一些病患並不適用，因這些等級為使用長腿支架 (**long leg brace**) 來行走或站立，而他們並沒有使用。巴歇爾指數天花板效應 (**ceiling effect**) 在緩慢進展的肌肉失養症病患非常顯著，而地板效應 (**floor effect**) 出現在裘馨型肌肉失養症病患。緩慢進展的肌肉失養症中以顏肩肱型肌肉失養症病患功能最佳，有比較好的下肢功能，日常生活功能也較好。本研究提供使用功能評估表於不同類型之肌肉失養症病患時之接受度，這些功能評估表出現的一些限制，在臨床上使用時應特別留意，尤其是使用於緩慢進展的肌肉失養症，建議使用時可考慮合併其他評估或使用題型結構較完整的評估量表。

關鍵詞：巴歇爾指數，功能狀態，肌肉失養症

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