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Case Report

Juvenile Muscular Amyotrophy of Distal Upper Limbs: A Case Report

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Juvenile muscular amyotrophy of distal upper limbs (Hirayama disease) is a rare disease predominantly affecting the anterior horn cells of the cervical cord in young men. It is a kind of cervical myelopathy characterized by insidious onset of unilateral distal dominant upper limbs muscle weakness and atrophy due to anterior cervical cord compression. It is difficult to differentiate this disease from other diseases with similar symptoms such as motor neuron disease. Cervical magnetic resonance (MR) study in flexed position is helpful to confirm the diagnosis.

We reported a 21-year-old man who complained of slowly progressed muscle atrophy and weakness of right hand and forearm after right shoulder stretching injury three years ago. Electrodiagnostic study revealed acute and chronic denervative change in the right side atrophied muscles. Routine cervical MR images showed high signal intensity over the anterior horn cells of the lower cervical cord. With the suspicion of Hirayama disease, flexion MR was performed and the striking and pathognomonic picture of anterior shifting of posterior dura at the lower cervical spinal canal was noted. He received neck collar therapy and vitamin B12 supplement. No further progression of symptoms was noted at the 3 months follow-up study.

Though Hirayama disease is spontaneous arrested, early diagnosis is necessary because early cervical collar application by preventing neck flexion has been shown to stop disease progression. Surgical managements are preserved to late stage. In cases of early onset of distal upper limb weakness with cold paresis and contractile fasciculation, the finding of asymmetric lower cervical cord atrophy on routine cervical MR study raise the suspicion of Hirayama disease. A flexion MR study should be performed to confirm the diagnosis. (Tw J Phys Med Rehabil 2010; 38(2): 107 - 114)

Key Words: Hirayama disease, juvenile muscular atrophy, monomelic amyotrophy

INTRODUCTION

Hirayama disease, also termed brachial monomelic amyotrophy, benign focal amyotrophy or juvenile muscular atrophy of distal upper limbs,[1,2] is a focal motor neuron disease affecting primarily young men in their teens and early twenties.[3] This disease has been reported chiefly from Asia and few from the western countries. [1,4] Characteristic presentations include insidious onset of

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weakness and atrophy in one or both upper limbs commencing in the distal upper limbs, followed by a spontaneous arrest within several years.[3,5] A flexion magnetic resonance (MR) study is warranted to prove this diagnosis.[5-10] Early diagnosis is important because early application of a cervical collar to minimize neck flexion may prevent disease progression.[11] Surgical managements are preserved for severe cases.[12,13]

We reported a case of Hirayama disease and reviewed the clinical presentation, image finding and treatment about this disease in order to help clinicians in diagnosis and treatment of this disease.

CASE REPORT

A 21-year-old right-handed man had a 3-year history of slowly progressive weakness and atrophy of his right hand and forearm after an episode of stretching injury of right shoulder. More recently, he noted aggravated weakness of the right hand when he flexed his neck. Weakness of fingers also exacerbated in cold weather (cold paresis). He only has skin atopy history and no other systemic disease or previous head or neck trauma history. His family history was non-contributory. His growth burst age was junior high school age. His leisure activity was playing basketball.

Neurological examination showed significant atrophy of muscles in his right forearm and hand including the flexor carpi radialis (FCR), thenar, hypothenar, and interosseous muscles (Table 1). Fine, irregular and asynchronous tremors of the right fingers were also noted while extending fingers (contractile fasciculation). The left hand presented a similar picture, but was less severe than on the right hand. Manual muscle strength of right wrist extension and elbow extension was graded as 4/5 and finger grasp and abduction was graded as 3/5. (Table 1) The deep tendon reflexes of right upper limb were decreased. No ataxia, extrapyramidal signs, Babinski sign, or abnormalities in sweating and urination were noted. Sensation to pinprick, vibration, and joint position was intact. Complete blood count and electrolyte were normal except high eosinophil level (11.4%, normal: <5%).

Electromyography showed active and chonic denervation in right FCR, triceps, first dorsal interosseus and bilateral abductor pollicis brevis muscles. Nerve conductive study showed normal conduction velocities of the median and ulnar nerves, but reduced compound muscle action potential (CMAP) amplitude of right ulnar nerve and decreased F-wave persistency in right median (31%) and ulnar nerves (31%). (Table 2 and 3) These findings were compatible with an anterior horn cell disorder involving right C6-T1 and left C8-T1 segments of the cord.

The plain cervical spine radiographs revealed no abnormalities except loss of physiologic lordotic curve. The cervical MR image in the neutral position revealed high signal intensity on T2-weighted image near the bilateral side anterior horns around C4 and C5 vertebral levels. In suspicion of Hirayama disease, a flexion cervical MR study was performed. MR images in flexion position showed ventrally shifting of the posterior dura and posterior spinal epidural venous dilation from C3 to C6 vertebral levels caused anterior cord compression at C4 and C5 vertebral levels (Figure 1A, 2A and C) Post contrast study shows enhancement of the posterior dural space. (Figure 1B, 2B and D) The diagnosis of Hirayama disease was confirmed. Neck collar was placed to prevent neck flexion and vitamin B12 was prescribed. No further progressive muscle atrophy was noted at the 3 months follow-up study with increased muscle strength. Cold paresis and contractile fasciculation also subjectively improved (Table 1). Electrodiagnositic studies showed reinnervation pattern (Table 2 and 3).

DISCUSSION

Hirayama disease develops predominantly in young men and more frequently involves upper limb with a period of progression of 3-5 years.[3,4,14] The etiology is hypothesized as disproportionate growth between the vertebral column and the contents of the spinal canal during the juvenile growth, which causes anterior cervical cord compression related to flexion movement of neck.[2,5,15,16] The onset age is approximately 2 years later than the peak age of the normal growth curve.[3,15] The mean onset age in Taiwan was at 16.8 years.[17] In our case, the onset age is 18 years, which is 2 to 3 years later than his peak of growth curve.

Table 1. Comparison of physical examination and lab data

	Pre-treatment	Follow up
Eosinophil count (%)	11.4	NA
Manual muscle power	R/L	R/L
Elbow flexion	5/ 5	5/ 5
Wrist extension	4/ 5	4-5/ 5
Elbow extension	4/ 5	4-5/ 5
Finger grasp	3/ 5	4/ 5
Finger abduction	3/ 5	4/ 5
Lower extremities	5/5	5/5
Forearm girth (cm)	R/L	R/L
Wrist up 6 cm	15/16	15/16.5
Wrist up 17 cm	21/25	21/24
Cold paresis	+	+/-
Contractile fasciculation	+	+/-

R: right side, L: left side, NA: not available

Table 2. Comparison data of nerve conductive study

Motor Nerves		Lat. (ms) R/L	Amp. (mV) R/L	CV (m/s) R/L	F lat. (ms) R/L
Median Wrist	Pre	4.0/3.5	10.1/9.4	NA	30.2/24.6
	F/u	3.4/NA	9.3/NA	NA	25.8/NA
Elbow	Pre	8.8/7.8	10.0/8.7	52/58	
	F/u	6.8/NA	8.7/NA	60/NA	
Ulnar Wrist	Pre	3.4/2.6	5.3/14.2	NA	27.2/25.2
	F/u	3.3/2.6	2.2/11.5	NA	25.4/24.7
Below Elbow	Pre	6.7/5.8	5.4/12.6	61/63	
	F/u	5.7/5.3	2.3/10.6	73/63	
Above Elbow	Pre	8.3/7.2	5.6/12.5	63/71	
	F/u	7.5/7.1	2.0/10.4	56/56	
Musculocutaneous	Pre	4.9/5.0	10.9/10.1	NA	
Axillary	Pre	3.5/3.1	17.2/17.1	NA	
Sensory Nerves:			(μV)		
Median Palm	Pre	0.8/0.6	57/58	66/78	
	F/u	1.9/NA	75/NA	64/NA	
Wrist	Pre	2.2/2.1	45/47	56/55	
	F/u	3.0/NA	79/NA	70/NA	
Ulnar	Pre	2.0/2.0	37/47	60/60	
	F/u	3.0/3.1	60/53	74/63	
LABC	Pre	1.4/1.5	10/8	NA	
MABC	Pre	2.5/2.2	22/12	NA	

R/L: right side/ left side, Lat.: latency, Amp.: amplitude, CV: conductive velocity, Pre: pretreatment, F/u: follow-up, LABC: lateral antebrachial cutaneous nerve, MABC: medial antebrachial cutaneous nerve, NA: not available

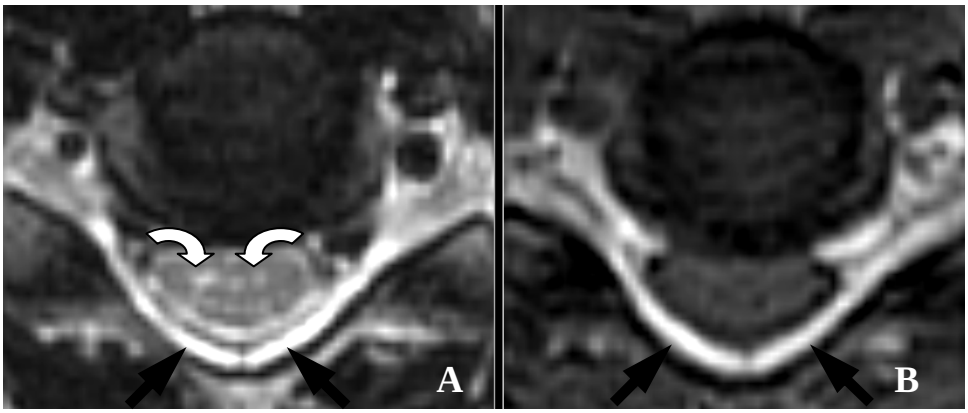

Figure 1. Flexion axial MR images at C4 to C5 disc level in a 21-year-old patient with progressive right-hand and forearm weakness and wasting for 3 years. A, T2-weighted (3300/114, repetition time/echo time) and B, contrast-enhanced (600/18) MR images show hyperintense signal near the bilateral of the side anterior horns (curved arrows in A) and enhancement of engorged posterior epidural space (arrows in A and B).

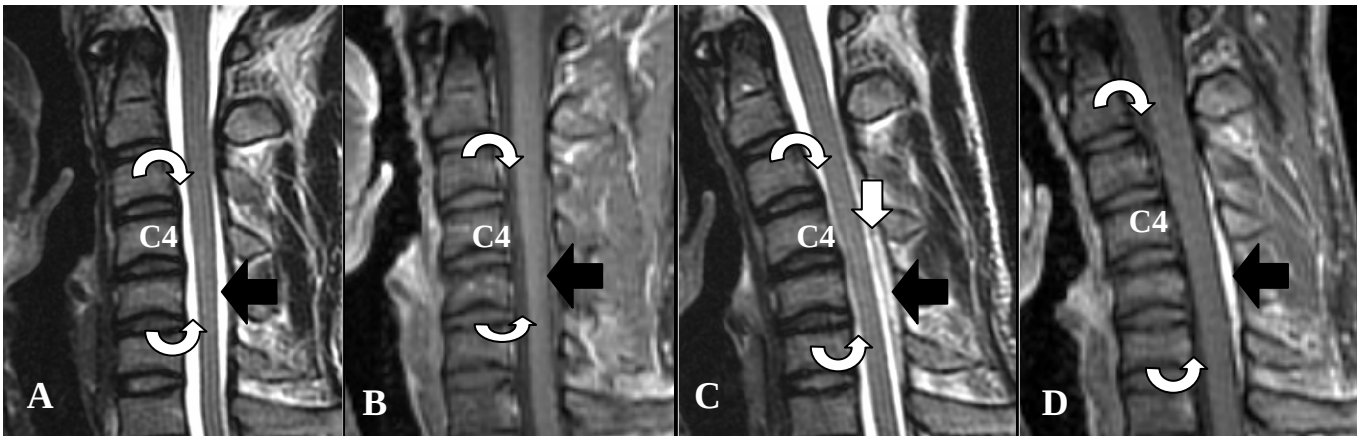

Figure 2. Cervical MR images. A, T2-weighted (3300/114) and B, contrast-enhanced (600/18) nonflexion sagittal MR images show wide subarachnoid space ventral to the spinal cord at C4 and C5 vertebral levels (curved arrows in A and B). The posterior dura is closely contact with spinal canal (black arrows in A and B). C, T2-weighted (5550/99) and D, contrast-enhanced (617/20) flexion MR images show forward displacement of tightened dural sac (white arrow in C) and the large posterior epidural plexus with enhancement (black arrows in C and D) from C3 to C6 levels. The spinal cord is compressed anteriorly by the C4 and C5 vertebral levels and causes narrowing of anterior subarachnoid space (curved arrows in C and D).

Some studies reported that heavy physical activity might be a precipitating factor of Hirayama disease due to repeated subclinical cervical trauma.[7,14,17] Strenuous exercise of the arms in sports was frequently noted in Hirayama patients. Other studies reported atopy, hyper-IgEaemia and hypereosinophilic syndrome could also be contributing factors to Hirayama disease.[18-23] In our case, the patient participated frequently in basketball and an episode of right shoulder stretching injury occurred before onset of muscle symptoms. He also had atopic history of dermatitis and higher eosinophil count (11.4%, normal value: 5%). Here, it doesn't reach the criteria of hypereosinophilic syndrome.

Table 3. Comparison data of needle electromyography study

Muscle		IA	Spontaneous activity (R/L)			Configuration (R/L)			Recruit	Activation
		R/L	Fibs	PSW	Fasc	Amp.	Dur.	Poly	R/L	R/L
Biceps	Pre	NL/NL	0/0	0/0	0/0	NL/NL	NL/NL	+1/+1	NL/NL	NL/NL
BR	Pre	NL/NA	0/NA	0/NA	0/NA	NL/NA	NL/NA	0/NA	NL/NA	NL/NA
FCR	Pre	↑ /NL	+2/0	+2/0	CRD/0	NL/NL	NL/NL	+1/0	↓ ↓ /NL	↓ ↓ /NL
	F/u	NL/NA	0/NA	0/NA	0/NA	+1/NA	NL/NA	0/NA	↓ /NA	↓ /NA
Triceps	Pre	↑ /NA	+1/NA	+1/NA	0/NA	NL/NA	NL/NA	+1/NA	↓ /NA	↓ /NA
	F/u	NL/NA	0/NA	0/NA	0/NA	NL/NA	NL/NA	0/NA	NL/NA	NL/NA
FDI	Pre	↑ /NA	+2/0	+2/0	0/0	NL/NL	NL/NL	+2/+2	↓ ↓ /NL	↓ ↓ /NL
	F/u	NL/NA	+1/NA	+1/NA	0/NA	+1/NA	NL/NA	+2/NA	↓ /NA	↓ /NA
APB	Pre	↑ /NL	+1/+1	0/0	0/0	+1/+1	NL/NL	+1/+1	↓ ↓ / ↓	↓ ↓ / ↓
	F/u	NL/NA	0/NA	0/NA	0/NA	NL/NA	NL/NA	0/NA	NL/NA	NL/NA

R/L: right side/left side, IA: insertion activity, Fibs: fibrillation, PSW: positive sharp wave, Fasc: fasciculation, Amp: amplitude, Dur: duration, Poly: polyphasics, Recruit: recruitment pattern, Biceps: biceps brachii, BR: brachioradialis, FCR: flexor carpi radialis, Triceps: triceps brachii, FDI: first dorsal interosseous, APB: abductor pollicis brevis, Pre: pretreatment, F/u: follow-up, CRD: complex repetitive discharge, NL: normal level, NA: not available

The characteristic clinical features of Hirayama disease include: 1) distally dominant muscle weakness and atrophy in the forearm and hand (brachioradialis muscle is relatively spared);[11,24] 2) young age at onset (10 to the early 20's);[17] 3) unilaterally dominant symptoms and signs (bilateral involvement can be seen in severe cases);[2,24] 4) incidental onset with a stationary stage after a progressive course;[1,3,5] 5) lack of remarkable sensory disturbance, and no symptoms in the lower extremities; and 6) exclusion of other diseases (such as syringomyelia, spinal cord tumors, motor neuron disease, etc). [3,8,9] Other symptoms include cold paresis (weakness of fingers easily worsens on exposure to cold environment) and contractile fasciculation (fingers tremor on weak finger extension).[17,25] The deep tendon reflex should be normal in Hirayama disease.[17] In our case, the decreased deep tendon reflex of his right upper limb might be due to severe muscle atrophy.

Electrodiagnostic study of our case fits the typical presentation of Hirayama disease. Electromyography showed acute or chronic denervation of atrophied muscles.[2,3] Nerve conduction study showed reduced CMAP amplitude, with ulnar motor nerve affected more frequently than median nerve. The cause for this discrepancy is unclear, although both originated from same spinal segments. [17] F wave showed low persistency.[12] Sensory nerve study remained normal in most patients,[17,25,26] which localized lesion as proximal to dorsal root ganglion. Moreover, the follow-up electromyography showed regeneration pattern.[2]

Cervical spine x-ray of Hirayama disease often reveals normal or loss of physiological curve. Routine cervical MR images can exclude organic lesion such as syringomyelia and spinal cord tumors.[9,15] Some special findings of Hirayama disease in cervical MRI with neutral position have been reported, including loss of attachment between the posterior dural sac and subjacent lamina and atrophy of anterior horn. [5,8] The images in neutral position of our case only show high signal intensity over the anterior horn cells of bilateral sides. This finding fits the pathogenesis of acute ischemic change related to anterior cord compression.

The most diagnostic image study is cervical MRI with neck flexion.[5] Several studies reported the pathognomonic features of Hirayama disease in flexion MR studies. The flexion MR image may reveal anterior shift of the posterior dura sac, flattening of the lower cervical cord, posterior spinal epidural venous dilation and localized cord atrophy, which fit the description of cervical flexion myelopathy. [6,8-10] The image findings of our case

also demonstrated these features, except lower cervical cord atrophy.

The pathophysiology of repeat cervical trauma related to neck flexion inspires the treatment of neck collar therapy. Early cervical collar therapy induces a premature arrest and shortens duration of illness.[2,11] It's also helpful to minimize functional disability.[11] Surgical managements such as anterior cervical decompression and fusion have been reported beneficial for patient non-responsive to conservative treatment.[11] For advanced Hirayama disease, tendon transfer improved the activities of daily living.[12] In our case, the patient responded well to neck collar use and vitamin B12 supplement. No further muscle weakness was noted after conservative treatment. Cold paresis and contractile fasciculation also improved.

CONCLUSION

The physicians should keep in mind that in case of insidious onset of unilateral distal upper limb weakness in young man with cold paresis and contractile fasciculation, the finding of asymmetric lower cervical cord atrophy on routine MR studies is highly suggestive of Hirayama disease. A cervical flexion MR study should be performed to confirm the diagnosis. Early diagnosis is necessary because early cervical collar application to minimize neck flexion has been shown to prevent progressive muscular weakness and stop disease progression.

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青少年遠端上肢肌萎縮：病例報告

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平山症(Hirayama disease)為一罕見的疾病，主要影響年輕男性的頸髓前角細胞，其特徵為因頸部屈曲動作壓迫頸部前方脊髓，造成一種緩慢發生，且以單側上肢遠端肌肉無力及萎縮為主的頸髓病變。臨床上不易與如運動神經元疾病等，其它具類似症狀的疾病區分。頸部屈曲時的核磁共振影像有助確認診斷。

本病例報告一名 21 歲男性，在三年前右肩部拉扯性傷害後，開始出現進展性的右前臂及手部肌肉的無力及萎縮。肌電學檢查顯示，右側萎縮肌肉處出現急性及慢性的去神經變化，常規頸部核磁共振檢查顯示下頸髓的前角細胞信號強度增加。因懷疑為平山症，我們安排屈頸核磁共振檢查，發現典型的下頸髓腔後方硬腦膜前移，証實了平山症的診斷。病患開始穿戴頸圈，同時補充維它命 B12。治療三個月後的追蹤檢查顯示症狀沒有惡化。

雖然平山症為一自限性疾病，早期診斷卻非常重要，因早期使用頸圈避免頸部前屈動作可阻止病程進展。臨床上若遇有年輕男性因單側遠端上肢無力就診，伴有冷麻痺(cold paresis)及收縮性肌顫動(contractile fasciculation)現象，同時常規頸部核磁共振檢查顯示下頸髓非對稱性萎縮，應考慮平山症的可能，並安排屈頸核磁共振檢查以確定診斷。（台灣復健醫誌 2010；38(2)：107 - 114）

關鍵詞：平山症(Hirayama disease)，青少年肌萎縮(juvenile muscular atrophy)，單肢性肌萎縮(monomelic amyotrophy)

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