Case Report

A Case Report of Hirayama Disease

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Abstract

Hirayama disease is also known as juvenile muscular amyotrophy of distal upper limb. This type of condition is rare and difficult to differentiate this disease from other disease with similar symptoms such as motor neuron disease. Very few cases reported in Medical science and its management is not much proved. But in our case a 20-year-old male came to department with complaint of slowly progressive muscle atrophy and weakness in distal U/L of right side. This disease had confirmed by MRI of flexed cervical region. Patient was managed by similar protocol: Vitamin B12, exercise of hand, cervical collar and isometric neck muscle exercise. Got benefit to patient for cessation of progressive of disease. It is necessary to early diagnosis of Hirayama disease so that patient got early management. Surgical management is preserved to late stage. In case of similar symptoms of cold paresis, amyotrophy, weakness of distal upper limb then a flexion MRI study should be performed to confirm the diagnosis.

Keywords: Hirayama disease, Monomelic amytrophy, Juvenile muscular amyotrophy.

Introduction:

Hirayama disease is a rare disease predominantly affecting the anterior horn cells of the cervical cord in young persons. It is cervical myelopathy characterised by insidious onset of unilateral distal dominant upper limb muscle weakness and atrophy due to anterior cervical cord compression. It has similar symptoms such as motor neuron disease and it is confirmed by magnetic resonance imaging (MRI) in flexed position. Hiryama disease has been reported mainly from Asia and in small number from the western countries[1,2]. This disease, also called as juvenile muscular atrophy of distal upper limbs[1,3], brachial monomelic amytrophy, benign focal amytrophy, is a focal motor disease affecting mainly young people in teen age/early twenties[4]. There is characteristic presentation of weakness and atrophy in one or both upper limbs, followed by spontaneous arrest within several years[5]. Early diagnosis is important because early application of hard cervical collar to minimise neck flexion may prevent disease progression[6].

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Cite as:
Kumar Deepak, Gogia Virinder Singh. A case report of hirayama disease. *IJPMR June 2013*; Vol **24 (2):** 51-5.

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Received on 22/04/2013, Accepted on 04/10/2013

Case Report:

This is a case of Hirayama disease in 20 years old male presenting with complaint of weakness of right hand and forearm for more than one and half years on first visit. The weakness was of progressive in nature with muscle atrophy. Recently he had noticed aggravated weakness of right hand and forearm along with tingling and fasciculation in right ring and little fingers. There was no definite history of trauma except few uneventful childhood traumas which were not contributory to present condition. Weakness in grip and pain on medial side of elbow used to increase on exposure to cold (cold paresis). He did not have any past or concurrent history of any other systemic disease. His family history was also not contributory. Patient did not have addiction to any drugs, tobacco or alcohol. Growth history was within normal limit. His hobby was playing cricket and watching movies on TV. A psychological assessment and counselling of patient and his father was also done to address the compliance to rehabilitation management issues.

Neurological examination showed significant atrophy of

muscles on his right forearm and hand including thenar, hypothenar and introsseous muscles (Figs 1 & 2). There were fine irregular and asynchronous tremors in right fingers while extending the fingers (contractile fasciculation). Patient had also complaint of pain and tingling sensation in right little, ring and middle fingers associated with particular type of movement of neck. Manual muscles test of right upper limb showed weakness as compared to left upper limb (Table 1). Deep tendon reflexes of right upper limb were diminished. There was no ataxia, extra pyramidal signs, abnormalities in sweating and urination. Sensations (light touch, deep pressure, vibration, proprioception and pin-prick) were intact.

Laboratory investigation such as CBC, ESR, electrolyte serum Ca^{++}, PO_4^{++}, vitamin D_3 and alkaline phosphatase was within normal limit.

In plain x-ray no any significant abnormalities seen except loss of lordotic curve. MRI cervical spine showed on flexion anterior imaging anterior shift of the posterior dural sac which was noted in the C6-C7 region causing mild flattening of the cord with enhancing posterior epidural mass seen on the contrast image (Figs 3 & 4) which appears to be extending from C4 level till the dorsal spine (Fig 4). Thecal sac diameter at intervertebral disc levels is shown in C4-C5 and C6-C7 (Table 2).

There is EMG and NCS done after clinical examination. Median nerve compound muscle action potential (CMAPs) was reduced and distal latency, F wave latency within normal range. Conduction velocities in all nerve were within normal range (Table 3). Sensory NCS was normal in all nerves (Table 4). In EMG flexor digitorum indices shows active denervetion in the form of there is fasciculation and fibrillation present and chronic nerve denervetion present in the form of neurogenic changes in C7, C8, T1 in myotomes. Extensor digitorum indicis showed no spontaneous activity, there were large polyphasic motor units with reduced recruitment. EMG of C5, C6 myotomes, namely deltoid, biceps brachi and brachiradilalis was normal. This features were suggestive of Hirayama disease.

Discussion:

Hiryama disease mostly found in young adult male and it involves frequently upper extrimities[7,8]. Hypothesized aetiology is as disproportionate growth of vertebral column and content of spinal canal during his growth period. Disproportionate growth affects anterior cervical cord compression due to flexion of neck[9,10]. The mean age is after 15 to 20 years. The onset age is approximately 2 year later than the peak age of the normal growth curve. In our cases onset age is 18 years which is 1 year later than his peak growth.

There are characteristic features of Hiryama disease ie, weakness and atrophy seen in distal and dominant hand muscle (brachiloradialis muscle is relatively spared)[11], young age of onset (10 to teen age of years), unilaterally dominant hand symptoms (rarely B/L hand involvement), onset be stationary after slow progression in years, sensory involvement absent and no lower extremities involvement, no other disease like sringomyelia , spinal cord tumours, MND etc. In this case we found that asymmetry is one of the most characteristic finding of this disease. So adolescent onset of distal upper limb weakness, the finding of asymmetric cord atrophy of flexion MRI studies confirm the diagnosis[11]. The

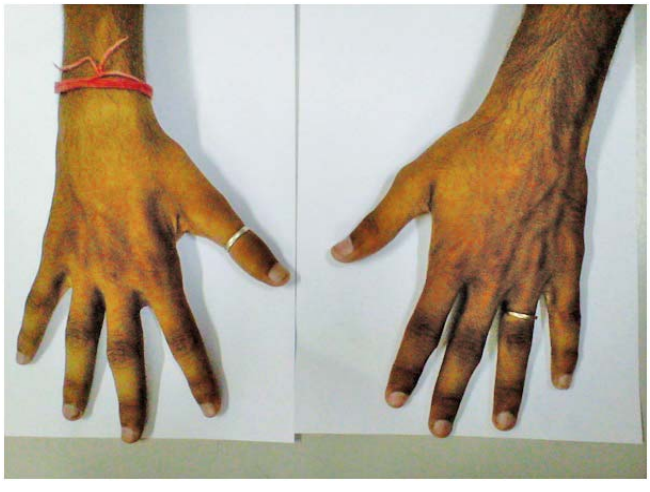

Fig 1- *Showing Significant Atrophy of Forearm Muscles*

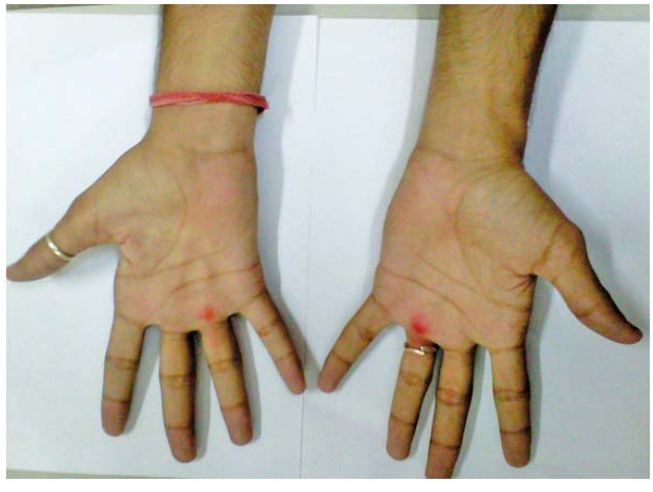

Fig 2- *Showing Atrophy of Thenar Muscles*

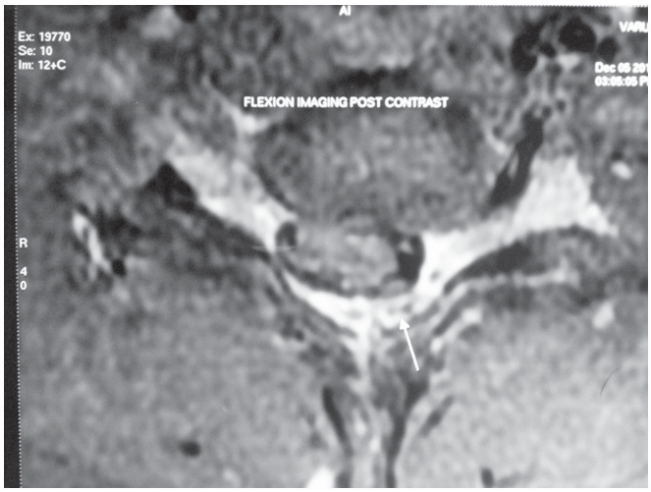

Fig 3- *Showing Posterior Epidural Mass in MRI*

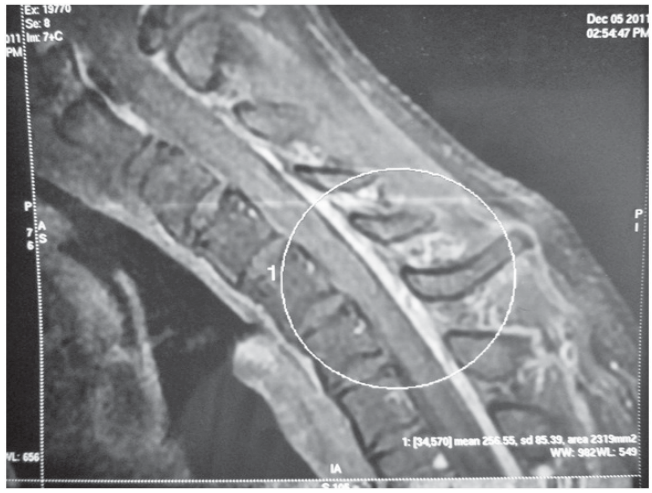

Fig 4- *Showing the Mass in MRI*

Table 1: *Manual Muscle Test*

Movement	Pre-treatment	Post-treatment (follow-up)
Elbow flexion	R4+; L5	R4+; L5
Elbow extension	R 4+; L5	R4+; L5
Wrist extension	R4+; L5	R4+; L5
Wrist flexion	R3+; L5	R3; L5
Supination	R4; L5	R4; L5
Pronation	R4; L5	R4; L5
Hand grip	R Poor; L Good	R Poor; L Good
Finger abduction	R3+; L5	R3+; L5
Finger adduction	R3+; L5	R3+; L5
Forearm girth	R 23cm L 26 cm	R 24cm; L 28 cm
Cold paresis	R Present; L Absent	R Present; L Absent
Contractile fasciculation	R Present; L Absent	R Present; L Absent
Static tremor	R Present; L Absent	R Present; L Absent

R: Right side; L: Left side; NA: Not available

pathophysiology of repeat cervical cord trauma (atrophy) due to neck flexion allow to prevent flexion of neck by hard cervical collar therapy. Early hard cervical collar therapy arrest the induces a premature arrest shorten duration of illness, also helpful to minimise functional disability[11]. Few cases which is not responsive to conservative treatment undergone to surgical treatment anterior cervical decompression and fusion got benefited[11]. For advance Hirayama disease, tendon transfer improves the activities of daily living[12]. In our case patient was advised for hard cervical collar therapy, vitamin B12 supplement along with strengthening exercise of right U/L and isometric neck muscle exercise. Patient has responded well and no further muscle weakness, cold paresis and fasciculation worsening seen.

Conclusion:

We should keep in our mind 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 MRI studies suggestive of Hirayama disease. There should be cervical flexion MRI study performed to confirm the diagnosis. Isometric neck muscle exercise is not proving for beneficial and requires more work over there. There should be early diagnosis because early use of hard cervical collar application to reduce neck flexion has

Table 2: *Thecal SAC Diameter at Intervetebral Disc Levels*

Intervetebral disc level	Antero-posterior diameter
C2-C3	12.0 mm
C3-C4	11.0 mm
C4-C5	10.0 mm
C5-C6	11.0 mm
C6-C7	10.0 mm

Table 3: *NCV (Nerve Conduction Velocity)*

Nerve and site	Latency	Amplitude	Segment	Latency difference	Distance	Conduction velocity
Median	**Nerve**	**Right**				
Wrist	4.0ms	5.7mV		ms	mm	m/s
Elbow	7.4ms	5.5mV	Wrist-Elbow	3.4ms	200mm	59m/s
Axilla	10.5ms	5.0mV	Elbow-Axilla	3.1ms	210mm	68m/s
Erb,s point	14.0ms	4.9mV	Axilla-Erb,s point	3.5ms	200mm	57m/s
Ulnar	**Nerve**	**Right**				
Wrist	3.3ms	4.7mV				
Below elbow	6.4ms	4.9mV	Wrist-Below elbow	3.1ms	170mm	55m/s
Above elbow	8.9ms	4.7mV	Below elbow-above elbow	2.5ms	110mm	44m/s
Erb,s point	14.7ms	3.1mV	Axilla-Erb,s point	3.5ms	180mm	51m/s
Median	**Nerve**	**Left**				
Wrist	3.7ms	9.5 mV				
Elbow	6.7ms	9.5mV	Wrist –Elbow	3.0ms	200mm	67m/s
Axilla	9.9ms	8.6mV	Elbow – Axilla	3.2ms	210mm	66m/s
Erb,s point	13.0ms	7.9mV	Axilla - Erb,s point	3.1ms	200mm	65m/s
Ulnar	**Nerve**	**Left**				
Wrist	2.9ms	9.1mV				
Below elbow	5.9ms	8.2mV	Wrist - Below elbow	3.0ms	170mm	57m/s
Above elbow	8.1ms	6.4mV	Below elbow - Above elbow	2.2ms	110mm	50m/s
Axilla	10.8ms	6.2mV	Above elbow - Axilla	2.7ms	150mm	56m/s
Erb,s point	13.8ms	5.7mV	Axilla – Erb's point	3.0ms	180mm	60m/s

Table 4: *Sensory Nerve Condition*

Nerve and site	Peak latency ms	Amplitude Micro volt	Segment	Latency difference Ms	Distance Mm	Conduction velocity m/s
Median	**Nerve**	**Right**				
Wrist	4.1	33	Index - wrist	2.5	150	60
Ulnar	**Nerve**	**Right**				
Wrist	2.8	24	Little finger - wrist	2.3	230	56
Median	**Nerve**	**Left**				
Wrist	3.4	39	Index - wrist	2.7	150	56
Ulnar	**Nerve**	**Left**				
Wrist	3.9	31	Short finger - wrist	2.5	130	52

been shown to prevent progressive muscular weakness and stop disease progression.

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