Case Report

DOI: 10.7860/JCDR/2015/13148.6374

Hirayama Disease: Escaping From the Quotidian Imaging

Radiology Section

ARJIT AGARWAL[1], SHRUTI CHANDAK[2], PAWAN JOON[3]

ABSTRACT

Hirayama disease is a rare type of neurological disease commonly manifesting as brachial monomelic amyotrophy in young males of Asian origin, easily understood as juvenile non-progressive cervical amyotrophy. The first case was reported by Hirayama in 1959. The pathogenesis is attributed towards chronic compression of cervical spinal cord during flexion movements of neck in cases where there is detachment of posterior dura mater. This chronic event, invariably leads to features of cord atrophy along with other MRI features. We report a case of 21-year-old male who presented with atrophy of distal muscles of his right hand and was sent for MRI of cervical spine which revealed prominent posterior epidural venous plexus without significant cord atrophy. Clinico-radiologic profile of the patient leads toward the diagnosis of Hirayama disease which was considered as borderline because of asymmetrical cord atrophy which is a not a routine imaging feature of the entity.

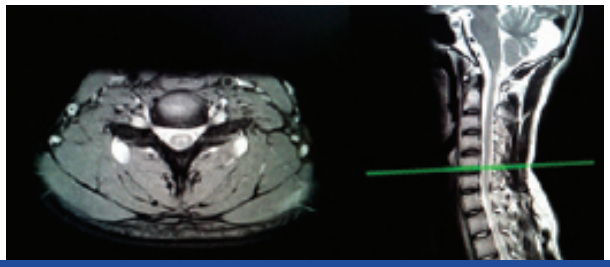
Keywords: Cervical cord, Dura mater, Monomelic amyotrophy

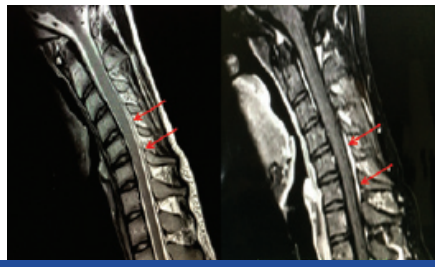
CASE REPORT

A 21-year-old young male presented to the Department of Neurology with weakness and atrophy of distal part of his right upper extremity involving predominantly the hand and forearm muscles compartment [Table/Fig-1]. The disease onset was from last one year with no significant progression in atrophy and weakness. There was no sensory or autonomic deficit, but on motor examination, he had grade 3 muscle weakness involving the forearm and hand with interossei muscles atrophy. Hyperactive deep tendon reflexes were present without fasciculations. Babinski sign was absent with normal cranial nerve examination. Minipolymyoclonus i.e. irregular coarse tremors were present in the fingers of the affected hand. Motor neuron disease and muscular dystrophy were kept as the differential diagnosis.

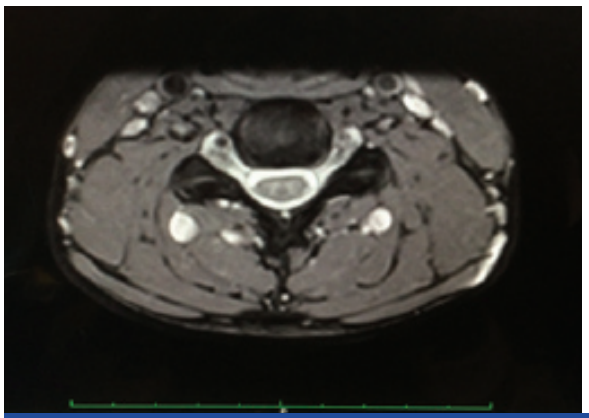
Routine laboratory investigations were within normal limits which included complete blood count, blood sugar, serum electrolytes, thyroid profile, renal and liver function tests. Patient also underwent nerve conduction study (NCS) and Electromyography (EMG) which demonstrated denervation potentials in the distribution of C7, C8 and T1 myotomes with reduced ulnar compound muscle action potential. There was no sensory involvement with normal C5, C6 distribution sparing biceps, deltoid and brachioradialis.

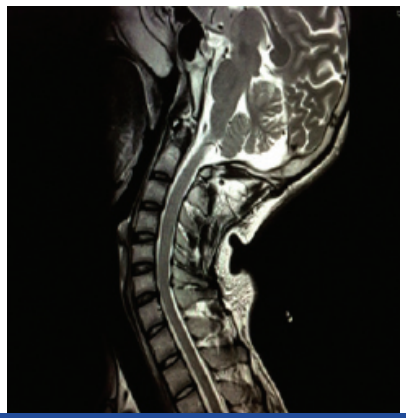
The patient was sent for dynamic MRI of cervical spine which was performed on 1.5T MRI (Siemens Magnetom Avanto 1.5 Tesla, Erlangen, Germany) in supine neutral, flexion and extension positions with contrast administration during flexion using Gadolinium based contrast agent i.e. Gadoversetamide (Optimark- 10.0ml; 500mM conc.). Imaging findings in neutral position were within normal limits except for the loss of cervical lordosis with maintained cord intensity and thickness [Table/Fig-2], while flexion MRI revealed anterior

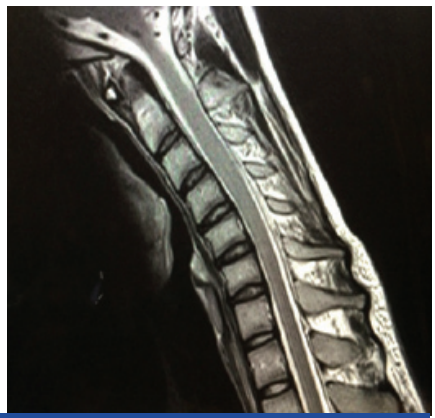




[Table/Fig-1]: Clinical photograph of the hand of the patient; showing atrophy of the distal muscles of the right upper limb with sparing of proximal muscle compartment **[Table/Fig-2]:** Neutral position Axial T2 weighted MRI showing normal cord intensity and caliber at the corresponding level in sagittal section **[Table/Fig-3]:** a) Flexion MR image of cervical spine on T2 weighted image shows detachment of the posterior dura mater from the lamina with prominent epidural flow voids (arrows); b) Post Gadolinium flexion MRI on T1 weighted image showing enhancing prominent epidural venous plexus (arrows)







[Table/Fig-4]: Flexion MRI axial T2 weighted image showing asymmetrical cord flattening on the right side **[Table/Fig-5]:** Extension T2 weighted MR showing repositioning of dura mater with effacement of posterior epidural space **[Table/Fig-6]:** Follow-up non contrast T2W flexion MRI of the same patient, taken after 1 year interval; showing similar prominent posterior epidural plexus at the same level

displacement of the dural sac with its detachment from subjacent lamina from C4 to C7 vertebral levels and prominent epidural flow voids which showed significant post contrast enhancement [Table/Fig-3]. Mild asymmetrical cord flattening was evident [Table/Fig-4], however; there was no significant localised cervical cord atrophy, a characteristic imaging manifestation from which this case of Hirayama disease is diverging. Normal repositioning of the dura was noted on imaging during neck flexion [Table/Fig-5].

The patient was diagnosed as a case of Hirayama disease, keeping the clinic-radiological profile in view and was advised to wear cervical collar. Follow-up scan was done after six months and one year interval; which revealed the same findings [Table/Fig-6] without any clinical progression.

DISCUSSION

Hirayama disease is an uncommon entity better known as juvenile flexion cervical myelopathy [1] or monomelic amyotrophy, presenting with weakness and atrophy of distal upper limb symmetrically or asymmetrically without sensory or autonomic function involvement. Clinical suspicion of the disease is much more common than its actual radiological confirmation because of its predominant occurrence in young males and with features masquerading that of motor neuron disease or spinal muscular dystrophy, however; the pathogenesis is completely different where it is more of embryological to anatomical origin. Hirayama disease is a benign entity predominantly occurring in the males during their second or third decades of life, with most of the cases being reported in the age group of 15 to 25 years [2]. There is initial progressive disease course which manifests as weakness, atrophy and infrequent cold paresis affecting the distal muscles of the upper extremity either symmetrically or asymmetrically, which later comes to a stand still. Pathological examination of the cervical cord revealed lesions in anterior horn cells from C5 to T1, more common and predominant at C7 and C8 levels [3]. The determining factor leading to involvement of anterior horn cells was disclosed by the current neuro-radiologic technique which shows the anterior displacement of the posterior dura from its subjacent lamina during neck flexion, chronically compressing the cervical spinal cord and leading to ischemic cord changes in the territory of anterior spinal artery which visually manifests in the form of cord atrophy, flattening, prominent enhancing epidural venous plexus and abnormal cervical curvature [1,2]. The growth imbalance between the vertebral column and the dural canal is the basic mechanism underlying the anterior displacement of the posterior dura [4] which causes laxity of the dura mater during extension (neck shortening) and tightening of the same on flexion (neck elongation). Male preponderance of Hirayama disease is attributable to the fact that there is differential growth between males and females [1].

Atopy was also considered as one of the predisposing factor in the pathogenesis of Hirayama disease as the incidence of atopic diseases and serum IgE was higher in these patients than in the unaffected population [4]. A study in Korean patients also identifies genetic association of monomelic amyotrophy with susceptibility genes like KIAA1377 and C5orf42 [5].

Imaging studies like conventional cervical spine X-ray and computed tomography (3D and dynamic) do not have any diagnostic role in Hirayama disease as the entity is not having any bony abnormality except altered cervical spine curvature which is commonly present in most of the routinely imaged patients. Dynamic flexion MRI of cervical spine shows loss of attachment of the posterior dura with cord atrophy [6,7] or asymmetrical cord flattening, and it is the imaging modality of choice [8]. Dilated epidural venous plexus is also striking and characteristic imaging feature which shows flow voids with enhancement on post contrast study. Neutral as well as the extension position leads to normal appearance due to repositioning of the dura by the mechanism described vide supra. Clinically suspected cases with posterior dural detachment, cord flattening and without cord atrophy are very scarcely reported in the available literature.

The disease progresses initially but comes to spontaneous arrest while the motor neuron disease progresses steadily [9], hence its early recognition is essential as it carries benign prognosis and fruitful outcome after effective treatments [10]. The disease is self limiting and conservative management with placement of a cervical collar is the treatment of choice to prevent neck flexion that will consequently prevent disease progression [11]. The other treatment option is surgical intervention in the form of anterior cervical decompression and fusion which is selectively advocated.

CONCLUSION

Clinical suspicion of Hirayama disease is part and parcel of the diagnosis as routine MR imaging protocols are inadequate to diagnose the disease where only relative segmental cord atrophy is a clue to further extend the imaging protocol.

REFERENCES

[1] Sonwalkar HA, Shah RS, Khan FK, Gupta AK, Bodhey NK, Vottath S, et al. Imaging features in Hirayama disease. *Neurol India*. 2008;56:22-26.
[2] Chen CJ, Chen CM, Wu CL, Ro LS, Chen ST, Lee TH. Hirayama Disease: MR Diagnosis. *AJNR Am J Neuroradiol*. 1998;19:365–68.
[3] Hirayama K, Tomonaga M, Kitano K, Yamada T, Kojima S, Arai K. Focal cervical poliopathy causing juvenile muscular atrophy of distal upper extremity: a pathological study. *J Neurol Neurosurg Psychiatry*. 1987;50:285-90.
[4] Kira J, Ochi H. Juvenile muscular atrophy of the distal upper limb (Hirayama disease) associated with atopy. *J Neurol Neurosurg Psychiatry*. 2001;70:798-801.
[5] Lim YM, Koh I, Park YM, Kim JJ, Kim DS, Kim HJ, et al. Exome sequencing identifes KIAA1377 and C5orf42 as susceptibility genes for monomelic amyotrophy. *Neuromuscul Disord*. 2012;22:394-400.
[6] Raval M, Kumari R, Dung-Dung AA, Guglani B, Gupta N, Gupta R. MRI findings in Hirayama disease. *Indian J Radiol Imaging*. 2010;20:245-49.
[7] Hassan KM, Sahni H, Jha A. Clinical and radiological profile of Hirayama disease: A flexion myelopathy due to tight cervical dural canal amenable to collar therapy *Ann Indian Acad Neurol.* 2012;15:106-12.
[8] Sarawagi R, Narayan S, Lakshmanan PM, Chakkalakkoombil SV. Hirayama Disease: Imaging Profile of Three Cases Emphasizing the Role of Flexion MRI. *J Clin Diagn Res*. 2014; 8(8):RD03-04.
[9] Hirayama K, Tokumaru Y. Cervical dural sac and spinal cord in juvenile muscular atrophy of distal upper extremity. *Neurology*. 2000;54:1922-26.
[10] Tashiro K, Kikuchi S, Itoyama Y, Tokumaru Y, Sobue G, Mukai E, et al. Nationwide survey of juvenile muscular atrophy of distal upper extremity (Hirayama disease) in Japan. *Amyotroph Lateral Scler*. 2006;7:38-45.
[11] Tokumaru Y, Hirayama K. A cervical collar therapy for non-progressive juvenile spinal muscular atrophy of the distal upper limb (Hirayama's disease). *Rinsho Shinkeigaku*. 1992;32:1102-06.

PARTICULARS OF CONTRIBUTORS:

1. Assistant Professor, Department of Radiodiagnosis, Teerthanker Mahaveer Medical College and Research Centre, Teerthanker Mahaveer University, Moradabad,(U.P.), India.
2. Assistant Professor, Department of Radiodiagnosis, Teerthanker Mahaveer Medical College and Research Centre, Teerthanker Mahaveer University, Moradabad,(U.P.), India.
3. Junior Resident, Department of Radiodiagnosis, Teerthanker Mahaveer Medical College and Research Centre, Teerthanker Mahaveer University, Moradabad,(U.P.), India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:

Dr. Arjit Agarwal,
A-46, Gandhi Nagar, Moradabad-244001 (U.P.), India.
E-mail: arjit123456@gmail.com

FINANCIAL OR OTHER COMPETING INTERESTS: None.

Date of Submission: **Jan 21, 2015**
Date of Peer Review: **May 12, 2015**
Date of Acceptance: **Jun 26, 2015**
Date of Publishing: **Aug 01, 2015**