Case Report

Bilateral familial Hirayama disease in a father and daughter

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Abstract

We are reporting a case of bilateral familial Hirayama disease where a father and daughter are the affected members of the family with the similar distribution of their weakness and wasting. To the best of our knowledge, bilateral familial Hirayama disease has not been described in father and daughter.

Key Words

Electromyography (EMG), Hirayama disease, magnetic resonance imaging (MRI)

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Sir,

Hirayama disease was first described by Keizo Hirayama as a juvenile muscular atrophy of unilateral upper extremity.[1] For many decades, the condition was thought to be unilateral and sporadic. Later on, reports of familial cases either isolated or part of a case series came up but the familial condition was rare and confined mainly to male members of family either father and son or to two brothers with the authors postulating autosomal dominant, autosomal recessive, and X-linked inheritance, but no genetic abnormality has been clearly discerned so far. Bilateral but asymmetrical cases were described by many authors as part of short or long case series but it was relatively uncommon compared to unilateral cases. Recently, bilateral symmetrical Hirayama cases have been described in a large Indian case series. The unifying feature of all these cases is the onset in late adolescence; pure motor weakness; and wasting of forearms and hand in C7, C8, and T1 myotomes with relative sparing of brachioradialis (oblique amyotrophy), and the dynamic magnetic resonance imaging (MRI) findings showing chronic neck flexion related ischaemic cervical myelopathy.[2]

We are reporting a case of bilateral familial Hirayama disease where a father and daughter are the affected members of the family with the similar distribution of their weakness and wasting. To the best of our knowledge, bilateral familial Hirayama disease has not been described in father and daughter.

Case Report

An 18-year-old girl had progressive weakness and wasting of left hand and forearm for 2 years which slowly progressed to involve right hand also. She also had action tremor of both hands (left >right) for a year. On examination, she had

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minipolymyoclonus (left >right) and asymmetrical wasting of bilateral forearm and hand (left >right) with relative sparing of brachioradialis (oblique atrophy) [see video]. On muscle power testing, there was weakness of long finger flexors and small muscles of the hand (left >right). There was a history of similar illness in father that started in the left hand at the age of 17 years and a year later involved both hands (left >right). His symptoms progressed for 4-5 years and then became stagnant without any specific treatment. Father had a more severe wasting of both forearms and hands with sparing of brachioradialis, diminished deep tendon reflexes in the upper extremities, and minipolymyoclonus in both hands [see video]. Sensory examination was normal in father and daughter. Their MRI of spine [see Figure 1] and electromyography (EMG) were suggestive of Hirayama disease. The amplitude of the compound motor action potential was reduced in father indicating more severe involvement [Table 1].

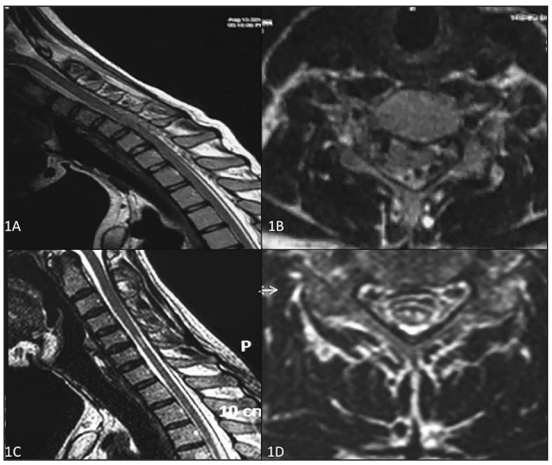

Figure 1: T2 weighted (T2W) magnetic resonance imaging (MRI) of cervicodorsal spine in neck flexion view:
Daughter: Sagittal (1A) and axial sections (1B)
There is an anterior displacement of posterior dura mater causing obliteration of the posterior subarachnoid space and compression of the spinal cord against C5, C6, and C7 vertebral bodies and prominent epidural flow voids extending from C4 to D1 vertebra (1A). There is an anteroposterior asymmetrical flattening (left > right) of the cervical spinal cord at C6 level, anterior displacement of posterior dura mater, and epidural flow voids (1B).
Father: Sagittal (1C) and axial sections (1D)
There is cord atrophy and a T2 hyperintense signal in the cord at C5 and C6 levels, note is made of epidural fat signal extending from C7 to visualised upper dorsal spine (1C). Corresponding T2 axial section at C5 and C6 levels, showed anteroposterior asymmetrical cord atrophy (left > right) and discrete hyperintense signals in the anterior segment of the spinal cord (1D).

Discussion

We are describing a unique case of bilateral familial Hirayama in a father and daughter with an onset in late adolescence, bilateral but asymmetrical (left >right) pure motor atrophy and weakness of forearm and hand with relative sparing of brachioradialis (oblique atrophy) and postural hand tremors.

Nalini *et al*.[3] in their 27-year experience of 190 cases of monomelic amyotrophy, reported a single familial case of a mother and son with unilateral affliction. The son presented at the age of 21 years with a 3-year history of unilateral weakness and wasting of left forearm and hand, which progressed for a year only, and the mother had her onset at the age of 20 years with the involvement of right forearm and hand and a disease progression of around 10 years. MRI of cervical spine was normal in the son but it showed myelopathic changes at C5 and C6 in the mother. In our case, the father's clinical condition has burnt out now whereas the condition in the daughter is still progressing. This is also depicted in their dynamic MRI of cervical spine which in the daughter showed a dynamic compression of the lower cervical cord during neck flexion while the one of father showed cord atrophy and a marrow signal change in the lower anterior cervical cord in neutral, flexion, and extension sequences. So the father's radiological picture closely matches with the mother whose case was reported by Nalini *et al*. suggesting that dynamic changes may be no longer found once the disease reaches a stationary phase. Misra *et al*.[4] (2005) reported the cases of 15 male patients with Hirayama disease from 14 Indian families. The condition had a unilateral onset in all but became bilateral subsequently in 8 patients. The condition was familial in two brothers. Sobue *et al*.[5] (1978) reported one family involving father and son in their series of 71 patients with Hirayama disease but no detailed description was available in the manuscript. Schlegel *et al*.[6] (1987) reported a case of a Caucasian man and his son with focal muscular atrophy of the upper limb having an onset at the age of 16 years in both. In the father, the disease started from right and then over the period of 1 year involved the left side as well, whereas in the son the disease started in the left forearm and hand and later on involved the right side with more severe affliction of left side. It was further supported by electrophysiological data, and disease was proposed to be due to the involvement of anterior horn cells from C5 to T1, but no radiological evidence was available. Tandan *et al*.[7] (1990) reported the case of identical male twins with chronic segmental spinal muscular atrophy confined to the upper extremities with age of 16 and 17 years, respectively, starting with right and then involved the left side. It was incidentally found in retrospective manner when one brother

Table 1: Median and ulnar nerve conduction velocity (NCV) and compound muscle action potential (CMAP) amplitude in daughter and father

Subject	Median (R) CMAP	Median (L) CMAP	Median (R) NCV	Median (L) NCV	Ulnar (R) CMAP	Ulnar (L) CMAP	Ulnar (R) NCV	Ulnar (L) NCV
Daughter	13.7	12.9	63.3	70.2	12.0	12.0	63.5	67.8
Father	10.4	7.4	52.5	55.3	2.4	2.7	58.2	45.4

*R = right, L = left, CMAP = compound muscle action potential, NCV= nerve conduction velocity

was admitted for some other indication at the age of 69 years. Authors described the clinical and electrophysiological findings well but MRI study was not available. In a large series of 106 patients of Hirayama disease by Pradhan *et al.*,[8] 11 patients had bilateral symmetrical distal upper limb involvement suggesting a severe degree of cervical myelopathy but none was familial.

Conclusion

To conclude bilateral Hirayama is an uncommon condition and familial Hirayama is a rare condition.

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Conflicts of interest
There are no conflicts of interest.

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