平山病1例

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1.概述

平山病为临床非常少见的一种良性自限性疾病，是一种与曲颈运动有关的颈髓病变。这是一种单纯的运动局灶性肌萎缩，主要累及 C7、C8 和 T1 脊神经及其支配的肌肉，该疾病不同于其他运动神经元疾病，主要区别为非进展性病程。目前，以颈托为主的保守治疗和手术治疗是治疗平山病的有效方法。

2.病例资料

患者，男，17 岁，因“进行性右手无力、肌肉萎缩半年”为主诉入院。既往体健，无神经系统疾病家族史。入院前 17 天右手表现出明显的无力及萎缩，随后第一次行 MRI 检查（距离发病约 5 月余）示未见明显异常。入院后神经系统检查：高级神经功能及颈神经功能正常。双上肢平举时震颤明显，右上肢远段肌力 4 级，近端肌力 5 级，左上肢及双下肢肌力 5 级，四肢肌张力正常，右手骨间肌及小鱼际肌萎缩明显，可见肌束颤动。四肢运动及感觉正常，双侧霍夫曼征及闭目难立征阴性，四肢腱反射减弱。

入院后完善辅助检查：肌电图显示双上肢明显的神经源性损伤特征，主要为左侧 C8 和右侧 C7–T1 水平支配肌失神经性损害。磁共振平扫（MRI）仅显示颈椎生理曲度变直，C5–6 椎间盘稍膨出（如图 A），而屈曲位磁共振平扫显示从 C3 到 T2 段椎管内后方硬膜外间隙广泛增宽（如图 B 和 C），病灶在 T1 加权像上呈长弧形等信号（如图 B），在 T2 加权像上呈现等信号和高信号（如图 C）。但是这种变化可能是由于积液或积血引起的，并导致相应的脊髓压迫，需要完善增强磁共振成像来进一步明确诊断。

简单地说，这个少年右手出现进行性无力、肌肉萎缩，特别是骨间肌和小鱼际肌。神经系统检查示四肢腱反射减弱，病理征阴性。整体来说，这些症状不符合近端肌无力的一般规律，因此肌病可能性不大，这可通过肌电图来证实。总之，这些症状符合神经源性病变，提示周围神经或下运动神经元病变，如周围神经病变、颈髓病变、脊髓肌肉萎缩症等。另外，颈髓病变包括脊髓型颈椎病、运动神经元病、平山病、脊髓空洞症、脊髓肿瘤等。

综上所述，该患者平山病可能性比较大，诊断依据该患者的临床表现、影像学和电生理学表现以及类似疾病的诊断要点，并且该患者颈椎磁共振有典型的“膜–壁分离”现象。这也符合文献报道的平山病的诊断标准。建议进一步完善增强磁共振检查以明确诊断，但患者家属拒绝。该患者在此期间病情稳定，因此我们根据患者家属意见给予保守治疗，如营养神经、佩戴颈托等治疗。

3.讨论

平山病为临床非常少见的一种良性自限性疾病，是一种与曲颈运动有关的颈髓病变。据报道，平山病为自限性疾病，通常进展缓慢，早期症状出现后常有 1–4 年的病程，经过此阶段，患者肌无力趋于保持稳定。目前，平山病的有效治疗方案是以颈托为主的保守治疗和手术治疗。

然而，对于周围神经损伤来说，主要症状是疼痛和感觉障碍，伴有肌肉无力和萎缩。脊髓型颈椎病主要表现为肌无力、萎缩及上肢上运动神经元受损症状，此多见于中老年人。此时，MRI 检查可以显示脊髓受压。运动神经元病可累及颈髓，随后逐渐出现下肢症状、锥体束征、球麻痹等。平山病是指青少年远端肌肉萎缩症，主要表现为肌肉无力、肌肉萎缩、肌束震颤及痉挛，通常发生在青少年时期[1–2]。平山病只会慢慢地影响上肢，而不会损害下肢、上运动神经元和脑神经。脊髓空洞症也可以表现为肌肉萎缩和手部小肌肉的肌束颤动，这可以发展到真性球麻痹，但进展缓慢，表现为有节段性分离性疼痛和体温下降，行 MRI 检查可看到空洞的形成。脊髓肿瘤有不同程度的传导束感觉障碍，但进展较快，行 MRI 和 CT 检查可发现占位病变[3–4]。

综上所述，平山病起病隐匿，虽然预后明显优于运动神经元病，但同样会造成患者一侧或双侧上肢永久性的肌肉萎缩，从而导致严重的功能障碍、劳动力丧失。但由于早期保守治疗和手术治疗有效，故早期诊断成为治疗平山病的关键，能阻止病情的进展，使患者获得较好的预后[5]。因此，尽早限制颈部过度屈曲，建议患者戴颈托 3–4 年，直到疾病自限。对于发病年龄小于 25 岁的上肢肌肉萎缩患者，应警惕平山病的可能性，积极行屈颈位 MRI 和神经电生理检查以明确诊断[5–6]。

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A case report of Hirayama disease

Luo huan，Zuo fangfang

Key words：Hirayama disease；motor neuron disease；muscular atrophy

1.Introduction

Hirayama disease is a benign self–limited disease that is rare in clinic，which is the cervical cord lesion related to cervical flexion. It is characterized by a pure motor focal amyotrophy in the distribution of C7，C8 and T1 spinal segmental–innervated muscles and differs from other motor neuron diseases by virtue of its ultimately non–progressive course[1–2]. At present，the conservative treatment mainly based on neck brace and the surgical treatment are effective for Hirayama disease[2].

2.Case Report

A previously healthy 17–year–old boy presented with progressive weakness and amyotrophia of the right hand in the recently six months. He had no family history of neurological diseases，but one day he was found obvious weakness and amyotrophia of the right hand seventeen days prior to admission. Then we acquired the first magnetic resonance image（MRI）more than 5 months after symptom onset，but there was no obvious abnormality. Upon neurologic examination，higher mental functions and cranial nerves were intact. The double upper limbs shaked slightly when stretched upward. Muscle strength was 4/5 distally and 5/5 proximally in the right upper limb and 5/5 throughout the left upper limb and the lower limbs. Meanwhile，there were amyotrophia in interossei and hypothenar

muscle of the right hand alongwith myofasciculi quiver. However, muscular tensions were normal in the limbs. And there were normal of the movements and sensations of the body including the temperature and vibration sensations. The Hoffmann sign and Romberg sign were negative. But the extremities tendon reflexes were weak.

And then the patient had some auxiliary examinations. The obvious features of neurogenic injury were found by needle electrode electromyography in the double upper limbs which showed the denervation damage of muscle activity innervated by the left C8 and the right C7–T1 spinal nerves. The magnetic resonance plain scan only showed the cervical vertebra physiology degree of curve became straightened and the C5–6 intervertebral disc was slightly distended (figure, A), while the flexion magnetic resonance plain scan showed the epidural space widened widely in the posterior spinal canal from the C3 to the T2 (figure, B and C). The lesion demonstrated a long arc equal–intensity on T1–weighted imaging (figure, B) .and equal– and hyper–intensity on T2–weighted imaging (figure, C). However, the change possibly caused by the hydrops or the hematocele and leaded the corresponding compression of the spinal cord. Enhanced magnetic resonance imaging was required to further clarify the diagnosis.

To make a long story short, this is a young boy with progressive weakness and amyotrophia of the right hand, notably the interossei and hypothenar muscle, on an insidious onset. The extremities tendon reflexes are weak, while the pathological signs are negative. Taken as a whole, the symptoms do not fit the general rules proximal weakness, therefore the myopathy is less likely proved through the electromyography. In a word, this is probably the neurogenic lesion which suggests a peripheral nerve or lower motor neuron lesion, such as the peripheral neuropath, cervical cord lesion, spinal muscular atrophy and so on. Moreover, the cervical cord lesion include the cervical spondylotic myelopathy, motor neuron disease, hirayama disease, syringomyelia, spinal cord neoplasms.

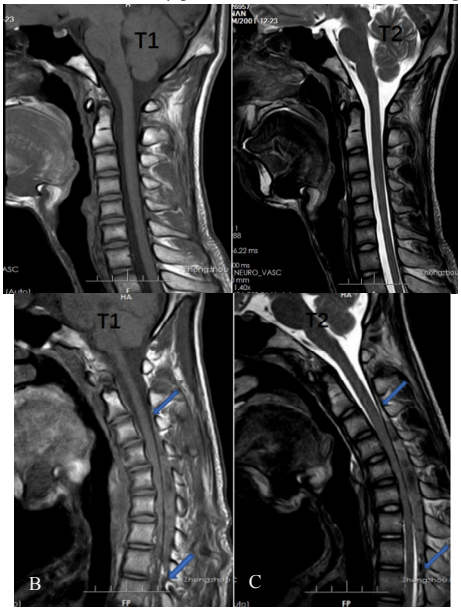
From what has been discussed above, this is probably the Hirayama disease according to the clinical manifestations, radiological and electrophysiological findings and the key points of diagnosis of similar diseases, and this patient have the typical "membrane–wall separation" on magnetic resonance image of the cervical spine. This also meets the diagnostic criteria of Hirayama disease reported in literatures[1–2]. Enhanced magnetic resonance examination is needed to further clarify diagnosis, but the family reject. This patient is stable during this time, so we give him the conservative treatment according to his family, such as nurturing nerve and wearing a neck brace.

3.Discussion

Hirayama disease is a benign self–limited disease that is rare in clinic, which is the cervical cord lesion related to cervical flexion. Hirayama disease is reported to be self–limited and usually progresses slowly, with a course of 1 to 4 years after the onset of early symptoms, then the patient's muscle weakness tends to remain stable. At present, the conservative treatment mainly based on neck brace and the surgical treatment are effective for Hirayama disease[1–2].

However, depending on the affected peripheral nerve, the main symptoms are pain and sensory impairment, accompanied with muscle weakness and atrophy. The cervical spondylotic myelopathy can present as muscular weakness and atrophy alongwith damaged to the upper motor neurons in the upper limbs, which usually occur in the middle and elderly aged people. In this way, the MRI examination can reveal compression of the spinal cord. The motor neuron disease can involve the cervical spinal cord and then gradually present with the symptoms of the lower limbs, pyramidal tract signs, bulbar palsy and so on. Hirayama disease, the young adult with distal extremity muscular atrophy, mainly present with muscle weakness, muscular atrophy, muscle bundle tremor and spasm, which often occur in the youth[1–2]. It only affects the upper limbs slowly, without damaged the lower limbs, the upper motor neuron and cerebral nerve. Syringomyelia can also present as muscular atrophy and myofasciculi quiver of the small muscles in hands, which can progress to the true bulbar palsy. It has segmental dissociative pain and temperature loss but progresses slowly. The MRI can see the formation of cavity. Spinal cord tumors have different degrees of conduction bundle sensory impairment but progresses relatively fast. The MRI and CT can see the space–occupying lesions[3–4].

In conclusion, Hirayama disease has insidious onset, and although the prognosis is significantly better than motor neuron disease, it can also cause permanent muscle atrophy in one or both upper limbs, leading to severe dysfunction and loss of labor force. While, as early conservative treatment and surgical treatment are effective, early diagnosis becomes the key to the treatment of Hirayama disease, which can prevent the progress of the disease and make patients obtain a better prognosis[5]. Therefore, excessive neck flexion is limited as early as possible and patients are advised to wear a neck brace for 3–4 years until the disease is self–limiting. So for patients with upper extremity muscular atrophy with the onset age less than 25 years old, the possibility of Hirayama disease should be vigilant, and the flexural MR and neuroelectrophysiological examination should be actively performed to make a definite diagnosis[5–6].



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