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Case Report

Hereditary spastic paraplegia-a differential for spastic paraplegia in children

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ABSTRACT

Hereditary spastic paraplegia (HSP) is a group of neurodegenerative disorders with familial origin mainly affecting the lower limbs. It has a prevalence of 3-10/100,000. The diagnosis is by symptomatology, clinical examination and neuroimaging to rule out other causes of spastic paraplegia. The diagnosis is confirmed by genetic analysis. Since this disorder is slowly progressive with nonspecific MRI brain findings the diagnosis can be delayed with delay in the start of rehabilitation measures. This disorder is usually diagnosed in the adult life and the literature has very few cases of paediatric HSP and hence we are reporting a case of HSP. Here we present the case of two siblings who presented to us with progressive weakness of both the lower limbs.

Keywords: Hereditary spastic paraplegia, Paediatrics, Neurodegenerative disorder

INTRODUCTION

Hereditary spastic paraplegia (Strumpell-Lorraine syndrome) is a rare heterogenous group of familial neurodegenerative disorders characterised by degeneration of the corticospinal tract and posterior column of the spinal cord. Clinically, they are classified as "pure" and "complicated "when other major clinical features are present.[1] Genetically autosomal dominant, autosomal recessive and X-linked forms are seen. Genetic analysis is necessary for confirmation of the diagnosis. MRI brain can help to exclude secondary causes of spastic paraplegia.

CASE REPORT

Here we present the case of two male siblings born to second degree consanguineous marriage who presented with progressive spastic gait disturbance noticed since the age of one year six months. The elder sibling is currently ten years old. He was brought with the history of difficulty in walking noticed since one and half years of age. When the child started to walk, he was noticed to have frequent falls and was bearing weight on the toes. It initially started in the left lower limbs and within 6 months was noticed to involve the right lower limb. As the child aged, he was noticed to drag his legs to help him walk and required support for walking. The child cannot climb stairs. There is no involvement of upper limbs, child is able to lift his arms above the head and is able use his hands for writing and mixing food without difficulty. There is no involvement of speech. Currently the child is able to walk with support, but has a spastic gait. On examination, the child has spasticity of bilateral lower limbs. The power in lower limbs was grade 2 in all muscle groups except for adductors and abductors of the hip which was grade 1. The deep tendon reflexes were symmetrically exaggerated in lower limbs with ankle clonus being present in both lower limbs. The plantar response was bilaterally extensor. The superficial reflexes were preserved. Upper limb motor system examination was normal except for deep tendon reflexes being

symmetrically brisk. The child had a spastic gait. He had also developed callosities over bilateral lateral malleoli. Sensory system, cerebellar system and autonomic system examination was within normal limits. Cardiovascular, respiratory and gastrointestinal system examination did not reveal any abnormalities.

The younger male sibling also had same complaints although in him the symptoms are more pronounced in terms that the child is unable to walk, but crawls to reach the destination. His examination also revealed the same signs as that of his brother.

The parents and the youngest sibling were examined and the examination did not reveal any deviation from the normal.

Clinical diagnosis of hereditary spastic paraplegia was made and MRI brain was done to rule out secondary causes for spastic paraplegia. MRI did not reveal any abnormality. Genetic analysis could not be done due to poor socio-economic status. The case was classified as "pure" form of HSP.

Figure 1: Image of the child showing walking with support.

Figure 2: Image of the child crawling due to spasticity.

Figure 3: Image of the child standing up with support.

DISCUSSION

HSP is a heterogenous group of heterogenous group of familial neurodegenerative disorders characterised by progressive lower limb spasticity. The prevalence is about 3-10/100,000.[2] HSP preferentially affect the corticospinal tract causing degeneration of longest axons. They are classified as "pure" form when spastic paraplegia occurs in isolation and "complicated" when associated with other neurological findings like seizures, dementia, amyotrophy, peripheral neuropathy, extrapyramidal symptoms.[3] The inheritance pattern of this condition can be autosomal dominant, autosomal recessive or X-linked. Each of these inheritance pattern can have the "pure" or "uncomplicated" forms. The low prevalence and insidious progression make it difficult for the physicians to diagnose and as a result the patients begin rehabilitation after many years of gait deterioration. HSP is usually diagnosed on a clinical basis after ruling out secondary causes of spastic paraplegia. MRI can reveal cortical, corpus callosal or cerebellar atrophy, white matter lesions, thinning cervical and thoracic cords but without any specificity.[4]

Genetic studies have revealed 31 different HSP loci.[5] Twenty HSP loci and nine HSP genes have been discovered. Out of these nine genes, five have been identified for autosomal dominant subtypes.[3,6] Mutations in the genes SPG4 and SPG3A account for up to 50% of all HSP cases.[7] These respectively encode for the protein's spastic and at last in. Mutations in the SPG4 is generally described as the pure form of the disease.

In our case, a clinical diagnosis of hereditary spastic paraplegia was made and it was diagnosed as "pure or uncomplicated" form on the basis of presence of isolated spasticity and absence of other neurological findings. Genetic analysis could not be done to poor socio-economic status.

CONCLUSION

Any patient presenting with progressive spasticity should elicit a differential diagnosis of HSP after ruling out secondary causes for spastic paraplegia. Earlier the

diagnosis, earlier would be the rehabilitation and can improve the quality of life of these patients.

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