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CASE REPORT: JUVENILE AMYOTROPHIC LATERAL SCLEROSIS: CLASSICAL WINE GLASS SIGN ON MRI

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ABSTRACT

Amyotrophic lateral sclerosis (ALS), also known as Lou Gehrig disease, is a chronic degenerative neurologic disease and is characterized by the selective involvement of the motor system. Usually patients present with upper motor neuron (UMN) and lower motor neuron (LMN) compromise. Degeneration of the UMN in the cerebral cortex is one of the main pathologic changes in ALS. These changes usually affects corticospinal tracts (CST) leading to degeneration of the fibres which show characteristic hyper intensities along the tracts leading to the "wine glass sign". Patients with ALS usually present in the 6th decade of life; presentation in pediatric age in the form of juvenile ALS (JALS) being rare.

INTRODUCTION

Amyotrophic lateral sclerosis (ALS) is a devastating neurodegenerative disease which affects both motor function and extra motor systems. According to the revised El Escorial criteria for the diagnosis of ALS the presence of signs for the affection of both upper motor neurons (UMN) in the primary motor cortex and lower motor neurons (LMN) in brain stem and spinal cord is mandatory, and the disease must be progressive [7]. Because of varied clinical phenotype of ALS, it is difficult to differentiate it from other ALS-mimicking conditions. Electromyography (EMG) can be used for the detection of LMN involvement in addition to the clinical examination. On the other hand, UMN signs must be visible at the clinical examination while electrophysiological transcranial motor stimulation (TMS) abnormalities are not included in making diagnosis of ALS according to the El Escorial criteria. Magnetic resonance imaging (MRI) is the radiological investigation of choice with the recent advances of diffusion tensor imaging (DTI), presenting a promising technique for early detection of alterations in the motor cortex and pyramidal tracts.

CASE REPORT

Herein we report a case of 9 year male who presented to the Department of Neurology with complaints of progressive increase in weakness of bilateral lower limb, poor grip in bilateral upper limb and difficulty in walking since last 2 years. There was no history of any muscle pain, trauma, fever, recent vaccination, dog bite, respiratory tract or gastrointestinal tract infection. Physical examination demonstrated decreased power in bilateral lower limb (3/5 in right lower limb and 2/5 in left lower limb) and upper limb (4/5 in right hand and 4/5 in left hand), no signs of dermal rashes or hypertrophy of muscles. Muscles found to be atrophied with exaggerated tendon reflexes and fasciculations. Laboratory findings including serum creatinine phosphokinase were within normal limits. No obvious pathological findings could be documented on plain computed tomography of the brain. Magnetic resonance imaging (MRI) of brain was performed using 1.5 Tesla scanner (GE Signa Excite, USA, 1.5 T). T2 weighted imaging and additional fluid attenuating inversion recovery sequence (FLAIR) showed hyperintense signals in posterior limb of internal capsule involving corticospinal tract(CST) which was found to extending upto midbrain. The coronal images showed classical wine glass sign.

DISCUSSION

Areas of abnormal signal intensity on T1 and T2-weighted images in the corticospinal tract have been previously reported in patients with ALS.[1-5] Other diseases such as ischemic processes, demyelinating diseases, infections, may involve the pyramidal tracts. However, bilateral symmetrical involvement associated with the characteristic clinical findings clinches the diagnosis of ALS.

The diagnosis of ALS was previously based only on clinical and electromyographic data. The degeneration of motor neurons may result in a cellular loss and axonal edema as previously shown on electronic microscopy.[6] So far, imaging methods had been of limited use because of their unsatisfactory results. Approximately 90% of

amyotrophic lateral sclerosis are sporadic with 10% having familial predisposition, but majority of the cases of JALS are familial.[7] About 20% of families with familial ALS carry mutation in superoxide dismutase gene (SOD1).[8] Most of juvenile ALS are autosomal recessive, though an autosomal dominant inheritance patterns have been described.[9] Recessive forms of juvenile ALS have been mapped to chromosome regions 2q33 and 15q12-21.[10] Genetic linkage to the chromosome 9q34 region is seen in the dominantly inherited form. No effective treatment for the disease have been found till date, though thyrotropin release hormone, ganglioside therapy and plasmapheresis have been tried in adults without much benefit.[9,11] Juvenile amyotrophic lateral sclerosis is a diagnosis that should suspected in a child presenting with a combination of upper and lower motor neuron signs. MRI facilitates reaching to the correct diagnosis by showing the typical involvement of the CST and the wine glass appearance.

CONCLUSION

JALS is an uncommon disease of the pediatric age group which has to kept as a differential in a child presenting with a combination of upper and lower motor neuron signs. MRI plays an important diagnostic tool for reaching up to a diagnosis and for prognosticating and in the follow up of the disease. Advancements in the field of structural and functional MRI techniques, a combination of DTI, resting fMRI and MRS might provide additional information of the cortical network failures especially amongst patients at risk. However, further validation studies are needed in form of meta- analysis to make these enter the routine MR imaging protocol.

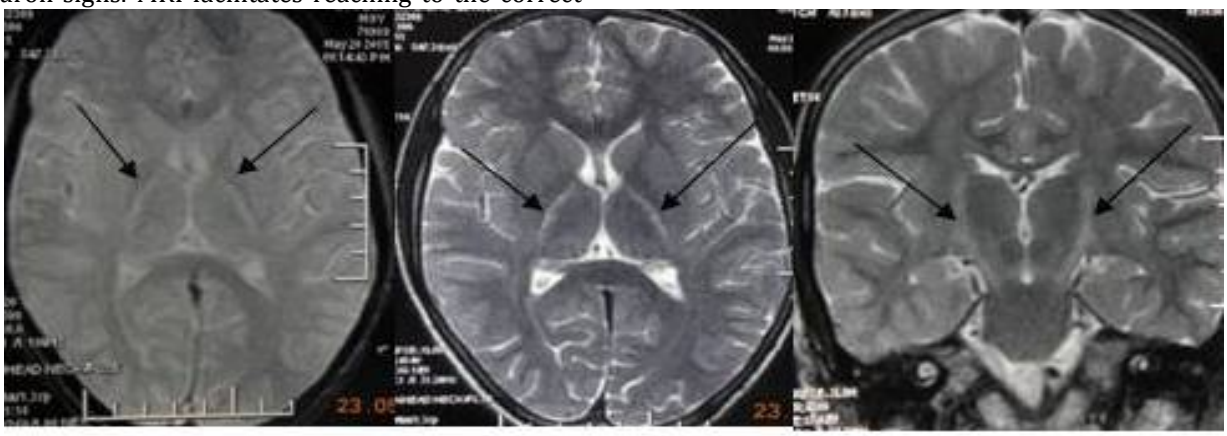

Fig.1 (a) Fig. 1(b) Fig. 1(c)

Fig. 1a: T1WI, black arrow shows hyperintensity in posterior limb of internal capsule
Fig. 1b: T2WI, black arrow shows hyperintensity in posterior limb of bilateral internal capsule
Fig. 1c: T2WI, black arrow shows hyperintesity in posterior bilateral internal capsule and mid brain (corticospinal tract) with classical wine glass appearance.

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