CASE STUDY

Metachromatic leukodystrophy: Magnetic Resonance imaging (diffusion weighted image - DWI)

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Abstract

Ten years old patient with juvenile form of metachromatic leukodystrophy (MDL) is presented. Apart from standard magnetic resonance (MR) protocol, the diffusion weighted sequence was performed and apparent diffusion coefficient (ADC) map was constructed. The cause of restricted diffusion is considered to be the deposits of metachromatic substance in the intercellular space, apparently similar reason for decrease of speed of diffusion in other storage diseases.

INTRODUCTION

Metachromatic leukodystrophy belongs to sfingolipidoses caused by deficiency of the enzyme arylsulfatase-A, resulting in the accumulation of sulfatide compounds in tissues and lysosomes [7]. It is autosomally recessive disease with impairment of growth or development of myelin sheath resulting in progressive demyelinization of the white matter. Three types of disease can be identified: infantile, juvenile and adult. Diagnosis is established on the basis of arylsulfatase-A activity in leucocytes and fibroblasts, increased content of sulfatides in urine and sometimes by biopsy of the sural nerve.

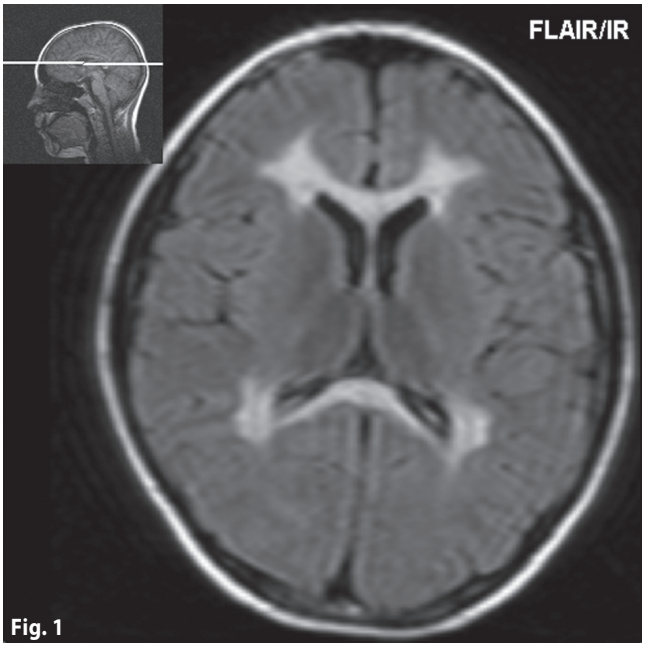
On MRI there are symmetrical lesions of decreased signal intensity on T1W images and increased signal intensity on T2W images located periventricularly, without any change after the administration of contrast agent. There are lesions in cerebellum, corpus callosum, posterior limb of the internal capsule. The U fibrous are affected in the late stage of the disease [1, 2].

CASE REPORT:

Ten years old patient with juvenile form of metachromatic leukodystrophy (MDL) is presented. The MRI study was performed on Philips (Intera) 1.5 T, in TSE (T2W), FLAIR, SE (T1W) and supplemented by contrast agent application Gd-DTPA. DWI with the use of echo-planar sequence (2918.28/75/ TR/TE, time acquisition 20s) (b=0 a b=1000mm^2/s). On automatically generated apparent diffusion coefficient (ADC) maps, the ADC values obtained in regions of interest were low (e.g. 0,55x10^{-3}mm^2/s) compared with the peripheral regions of the brain parenchyma (e.g. 0,95 $\times$ 10^{-3}mm^2/s).

MRI showed a symmetrical increase in signal intensity on T2W and on FLAIR sequences, moderate decrease in signal intensity on T1W sequence in areas of white matter periventricullary located without any change after the administering of contrast agent (Fig. 1, Fig. 2).

DWI showed restriction of diffusion (increased signal) with decreased signal on ADC map (Fig. 3).



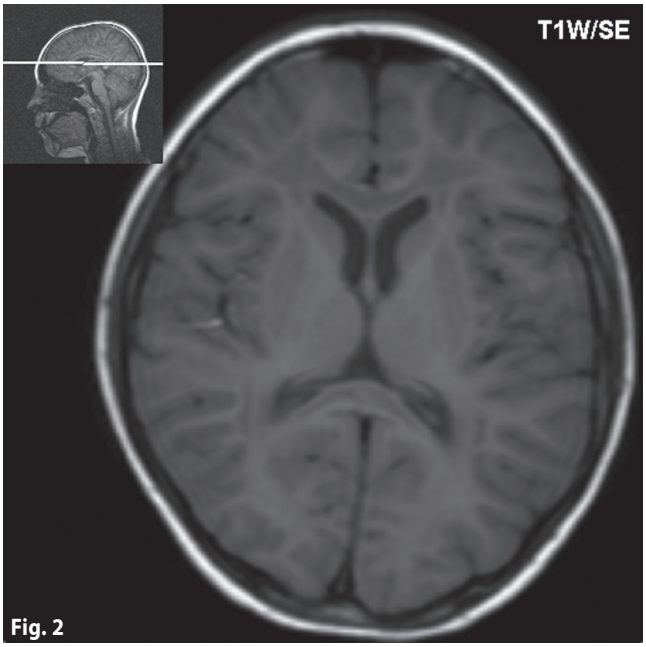
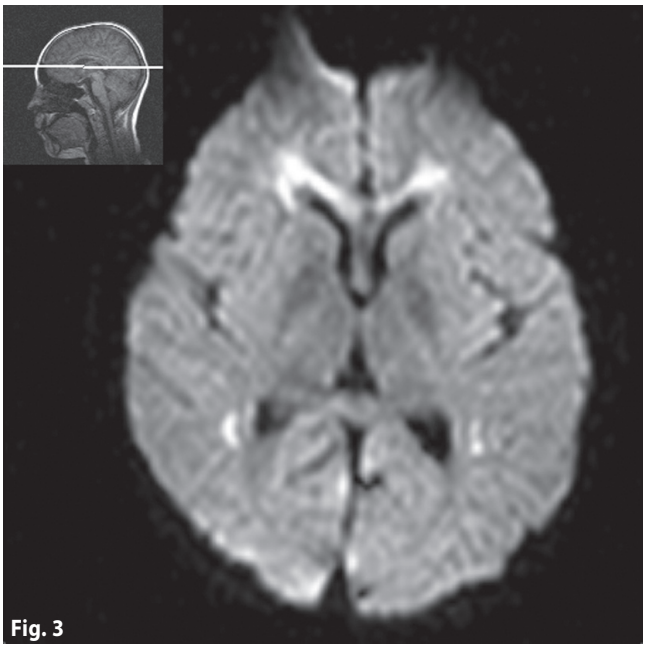


Figure 1: Transverse FLAIR. Increased signal intensity on FLAIR sequence in the areas of white matter, periventricullary located.

Figure 2: Transverse T1W image. Decrease in signal intensity on T1W sequence in the areas of white matter, periventricullary located.

Figure 3: DWI (b= 1000mm^2/s). Restricted diffusion can be seen in the same areas as in figures 1 and 2 (increased signal).



DISCUSSION

MDL is caused by a deficiency in the lysosomal enzyme sulfatide sulfatase (arylsulfatase A).

Some patients with clinical MLD have normal arylsulfatase A activity but lack an activator protein that is involved in sulfatide degradation. Both defects result in the accumulation of sulfatide compounds in neural and in nonneural tissue. In the neuronal tissue this impairs the development and growth of the myelin sheath and finally leads to demyelinization. This leads to proliferation of astrocytes (formation of scar tissue) with sparing of the demyelinated axons.

Demyelinization affects foremost periventricular white matter, but in latter stages of the disease, the demyelinization affects other structures of white matter as well (internal capsula, U-fibers…). In astrocytes and macrofages there are deposits of metachromatic granules with relative sparing of neurons and oligodendrocytes (their reduced numbers are noted in non affected tissue). Metachromatic material is seen in cellular cytoplasm under electron microscope (lysosomes) and in extracellular spaces under optic microscope [6]. There are deposits of pathological fats in pancreas, spleen, kidneys and even though the disease manifests itself primarily by neurological symptomatology, it is without doubt a systemic metabolic disease.

Some authors explain the restricted diffusion in DW MRI sequence by cytotoxic oedema [3, 5], similarly as is the case in acute stroke [8]. This assumption would imply the presence of cytotoxic oedema but in metachromatic leukodystrophy there are no signs of ischemia or cytotoxic oedema. Contrary to that there are deposits of metachromatic substances in cellular cytoplasma (lysosomes) noticeable under electron microscope and extracellular presence of metachromatic substances seen under light microscope [6].

We suggest that the deposits of pathological substance in extracellular space are the cause for decreased diffusion; this notion should also apply to a cause of restriction of diffusion in some other storage diseases.

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