Classical case of late-infantile form of metachromatic leukodystrophy

Sir,

We would like to share a rare case of late-infantile form of metachromatic leukodystrophy (MLD) presenting in our institution.

The parents of a 3-year-old boy reported in the pediatric outpatient department and gave a history of progressive psychomotor regression, disturbance of gait, and quadriplegia in their son for the last 1 year. The patient was admitted for workup, and magnetic resonance imaging (MRI) brain was performed the next day. The MRI scan revealed confluent T2 and fluid-attenuated inversion recovery hyperintensities in the periventricular white matter and centrum semiovale with linear and dot-like hypointensities within it characteristic of the “tigroid” and “leopard skin” appearance of demyelination [Figure 1a and b].

The posterior limb of internal capsule and the corpus callosum (genu, body, and splenium) were also involved [Figures 2 and 3]. The diffusion-weighted images revealed restriction in the periventricular white matter and corpus callosum [Figure 4]. The peripheral U fibers were spared, and no noticeable enhancement was seen in postcontrast images. In combination; the clinical and imaging findings were suggestive of late-infantile form of MLD. The diagnosis was confirmed biochemically by the reduced arylsulfatase A levels in peripheral white blood cells and 24 h urine assay.

MLD is a form of lysosomal storage disorder with autosomal recessive inheritance which occurs due to deficiency of arylsulfatase A enzyme resulting in accumulation of sulfatides in the peripheral and central white matter.[1] The prevalence of MLD is about 1 in
100,000 newborns\textsuperscript{[2]} and according to age, three forms have been described: Late-infantile, juvenile, and adult. Infantile form is the most common, manifesting at 12–18 months of age with regression of motor development, intellectual decline and disturbances of speech/gait, and blindness.\textsuperscript{[1,2]} MRI findings of MLD include symmetric confluent hyperintense areas in periventricular white matter with a demonstration of “tigroid” or “leopard skin” pattern in deep white matter.\textsuperscript{[3]} The radially oriented hypointense stripes or dots which give the tigroid/leopard skin appearance have been related to relative sparing of myelin in the perivenular region.\textsuperscript{[4]} Diffusion-weighted MR imaging reveals restricted pattern of cytotoxic edema in the affected white matter in the absence of ischemia, the cause of which is probably due to restricted mobility of water molecules in the abnormal myelin.\textsuperscript{[5]} Subcortical U-fiber sparing is characteristically seen in initial stage, but involvement may occur in later stages.\textsuperscript{[2]} There may also be variable involvement of corpus callosum, corticospinal tract, and cerebellar white matter.\textsuperscript{[1]} In postcontrast MR either no enhancement or punctate foci of enhancement can be seen in the background of nonenhanced demyelinated white matter.\textsuperscript{[1,2]}

Apart from MLD, tigroid appearance can also occur in Pelizaeus-Merzbacher disease, but differentiation is usually possible based on the clinical context and low levels of arylsulfatase A in the peripheral white blood cells and urine in the former.

Prognosis in MLD is not good with progressive quadriplegia, decerebration, and death within 6 months to 4 years after onset.\textsuperscript{[1]}

Our case is thus illustrative of the late-infantile form of MLD and showed typical clinical and MR imaging findings.
Letters to the Editor

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References


Sir,
Epilepsy is a chronic disorder of the brain occurring among people from all age groups across the world.¹ The disease has been acknowledged as a major global health concern owing to its high prevalence of 50 million and an incidence of 2.4 million each year.² From the socioeconomic perspective epilepsy has been associated with fear, myths and misconceptions, social stigma and discrimination to the patient and their family members, significant impact on the quality of life, loss of disability-adjusted life years, premature and sudden deaths, loss of work productivity, financial burden on the health care delivery system, and violation of human rights (viz., restricted access to health and life insurance, difficulty in obtaining a driving license, no eligibility for specific occupations, etc.).¹-³ Furthermore, in contrast to the estimates of annual new cases in high-income nations (viz., 30–50 people/0.1 million population), the incidence rates in middle- and low-income nations is almost double.² This is predominantly because of the high prevalence of infections (such as malaria, neurocysticercosis, etc.), road traffic accidents, birth trauma; weak health infrastructure, including shortcomings in health staffs and logistics; and limited accessibility to the desired package of service, in the developing nations.¹,² In addition, it has been estimated that almost 80% of the people with epilepsy are from low- and middle-income nations, of which 75% cannot avail the desired treatment (epilepsy treatment gap) due to various constraints.¹,² However, owing to the fact that close to three-fourth of people with epilepsy usually respond to anti-epileptic drugs, the affordable cost of annual medications, and definitive possibility to diagnose/treat at the primary level of health care itself, without the use of sophisticated equipments, it is an alarming concern that almost 75% of diseased people are still devoid of the treatment in low-resource settings.¹-³ There is an extensive need to improve public and professional awareness that epilepsy is a treatable brain disorder, enhance the level of acceptance with regard to epilepsy patients among the society,