

Metachromatic Leukodystrophy

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History

Early teen with new onset of difficulty in school and general regression.

Diagnosis

Metachromatic Leukodystrophy

Discussion

Metachromatic leukodystrophy is a rare autosomal recessive lysosomal storage disorder that results in a decrease of normal arylsulfatase-A or one of its cofactors. This results in failure of myelin breakdown with accumulation of ceramide sulfatide within macrophages and Schwann cells. This abnormal accumulation leads to demyelination.

There are three presentations depending on the age of the patient. The infantile form is the most common, presenting usually in the second year of life with ataxia and strabismus. Intellectual deterioration occurs gradually with these patients usually dying within 6 months to 4 years of symptom onset. The juvenile form often presents with declining school performance and death usually ensues within 20 years. The adult form may present with progressive corticospinal, corticobulbar, cerebellar, or extrapyramidal signs. Other organs that may be affected include the gallbladder, kidney, and liver.

Imaging findings include deep white matter changes of low attenuation on CT as well as T1 and T2 prolongation on MRI. There is often involvement of the corpus callosum, internal capsule and corticospinal tracts. A "tigroid" or "leopard" appearance of the demyelinated periventricular white matter is a characteristic thought to reflect sparing of the perivascular white matter. There is sparing of the subcortical U-fibers until very late in the disease. If there is cerebellar involvement, the white matter may appear hyperintense on T2. No enhancement is seen with contrast enhanced imaging. The differential diagnosis for the imaging appearance of metachromatic leukodystrophy includes Pelizaeus-Merzbacher disease, TORCH or pseudoTORCH infections, periventricular leukomalacia, Sneddon syndrome, Krabbe disease, and megalencephaly with leukoencephalopathy. Diagnosis is often confirmed by excess urine sulfatide or absent/deficient arylsulfatase A activity within leukocytes or fibroblasts.

Findings

Diffuse low density and abnormal intensity of the cerebral white matter without enhancement. Sparing of subcortical U fibers. Stripes of affected and unaffected myelin (tigroid appearance) on MRI. Corpus callosum atrophy and abnormal signal on MRI.

Reference

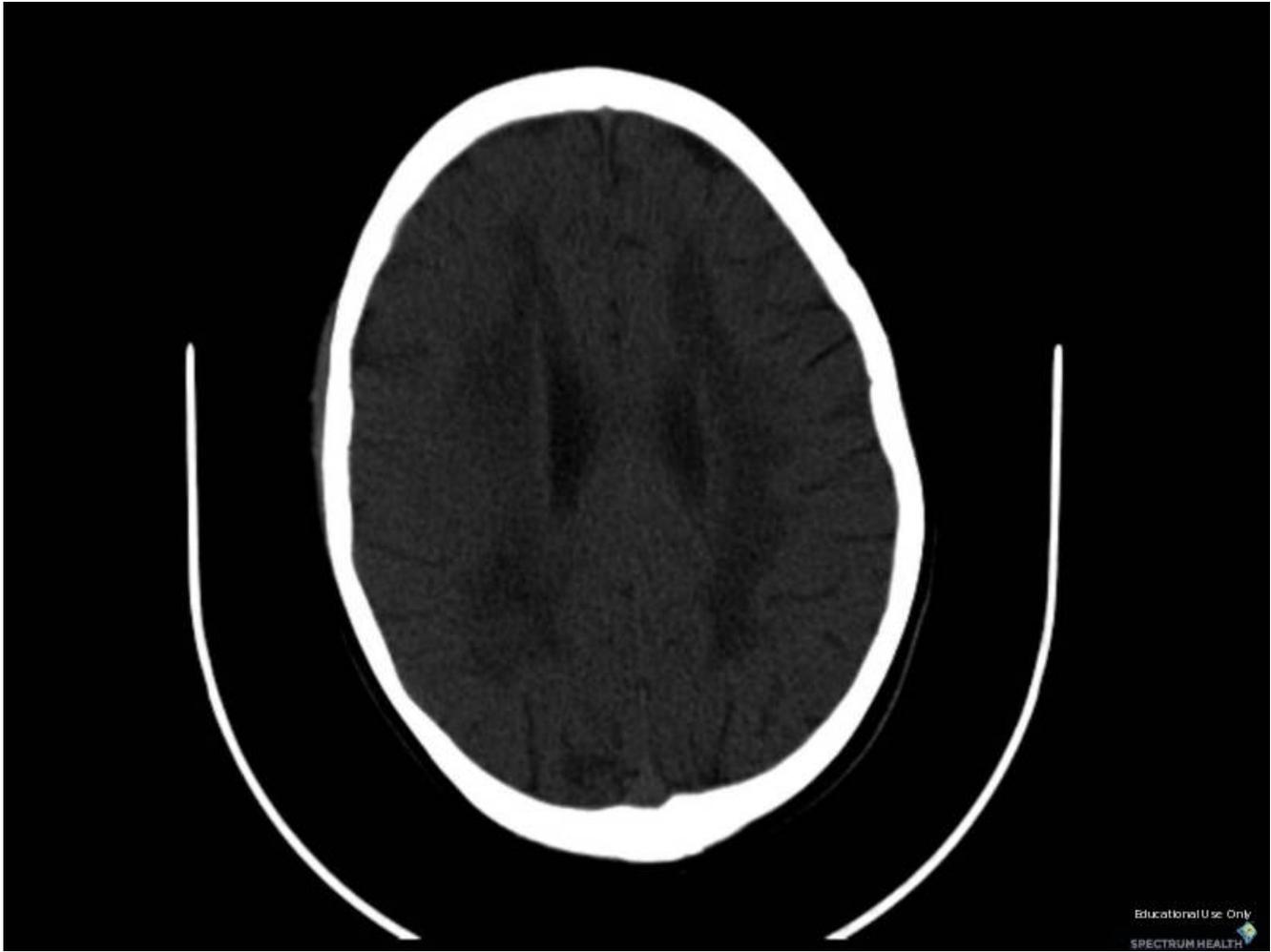
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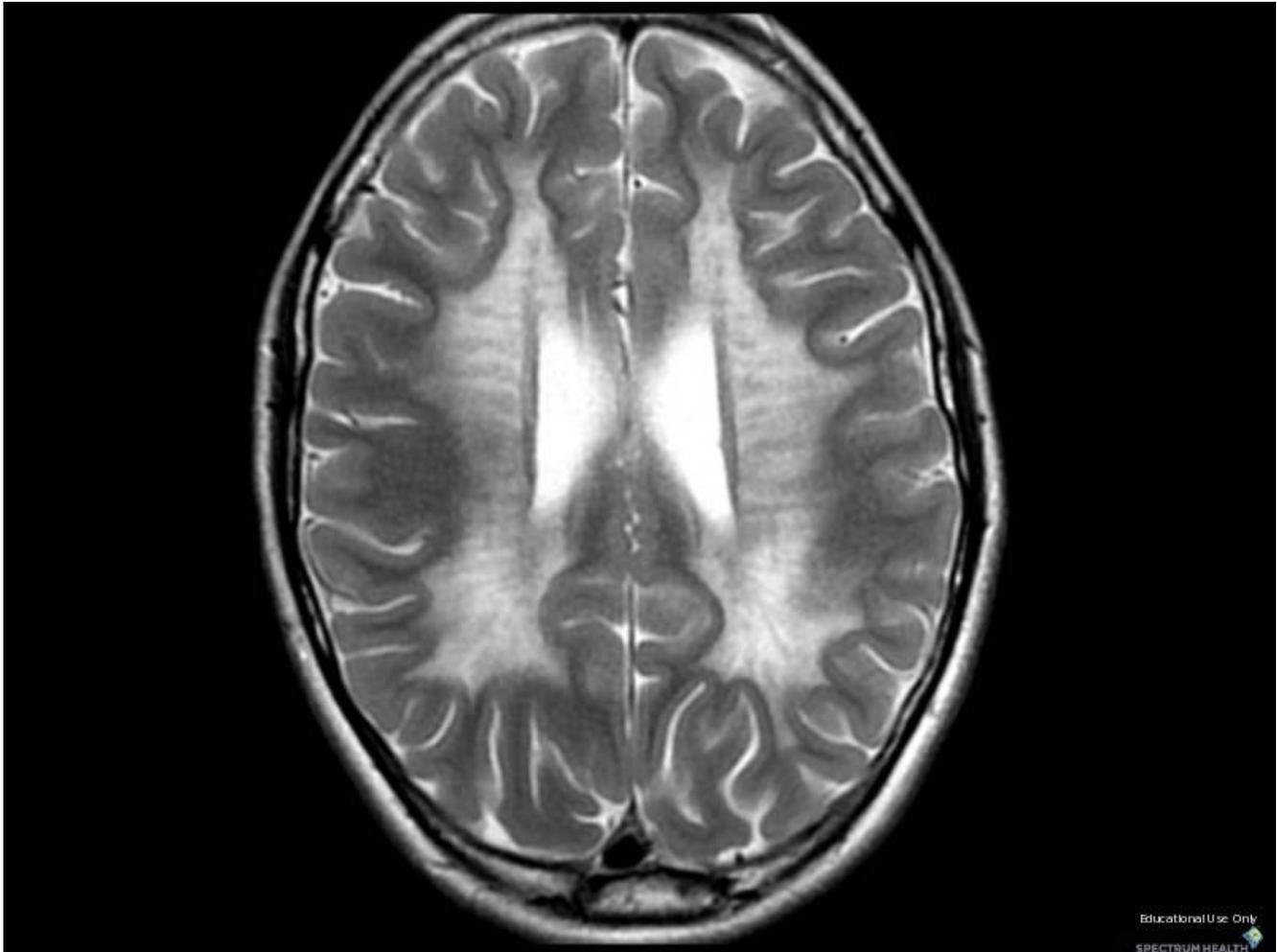
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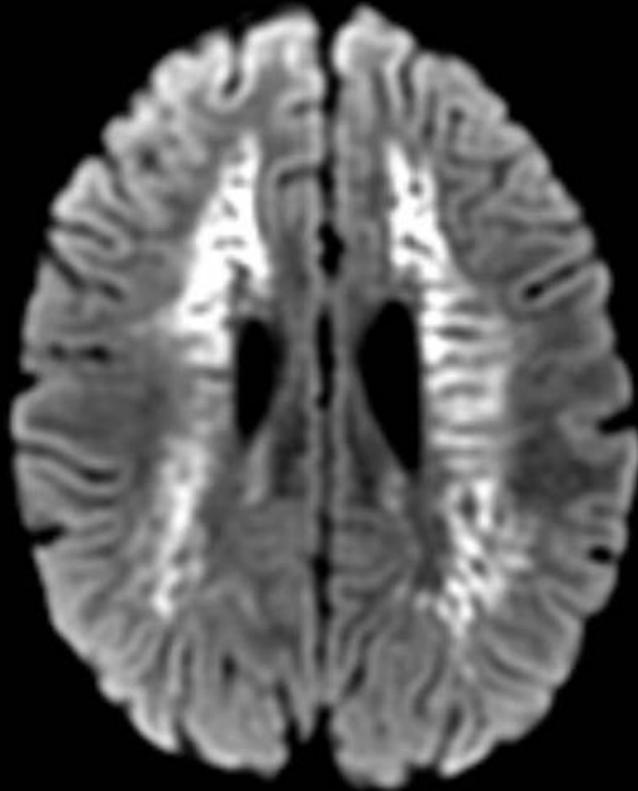
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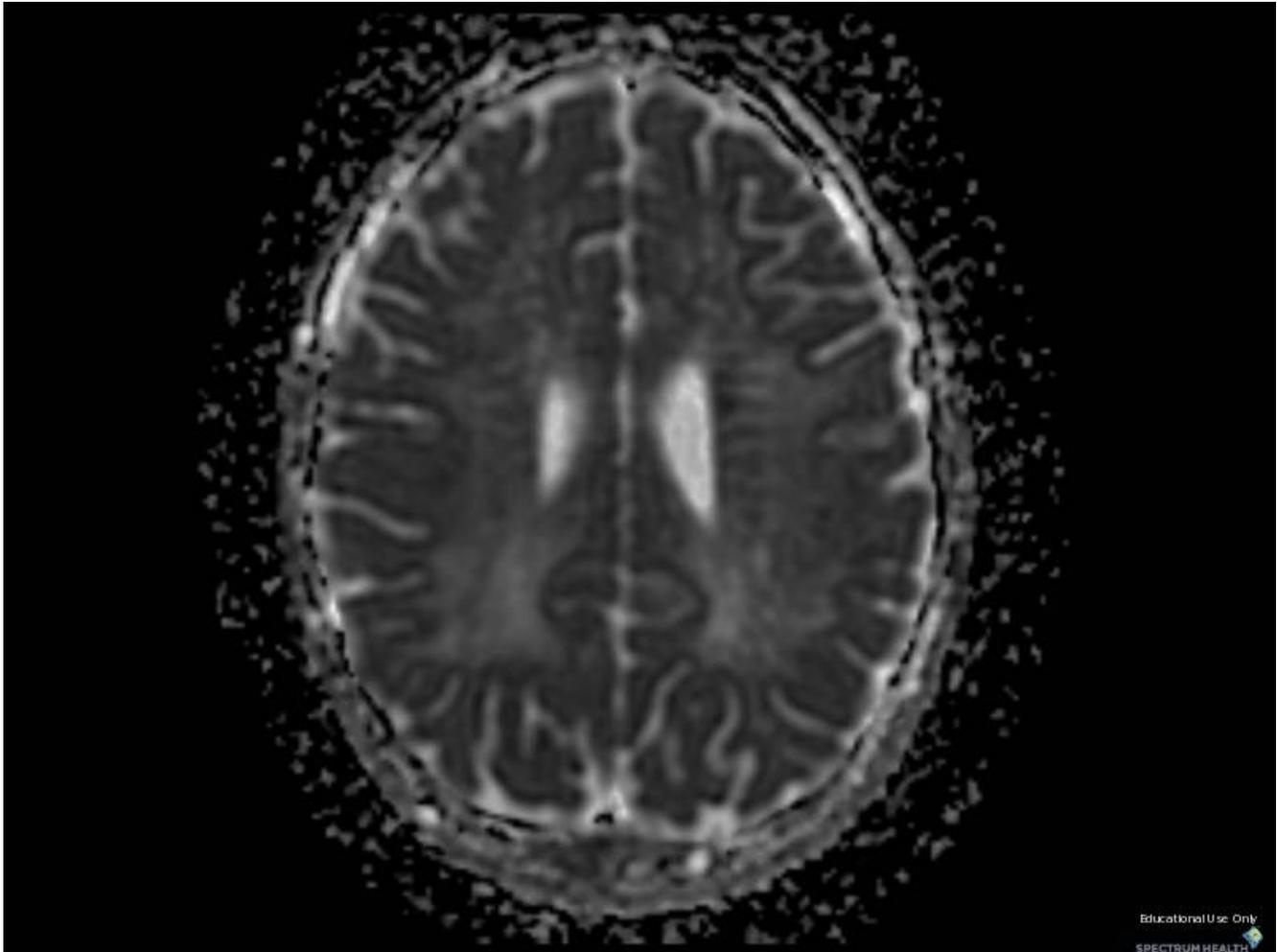
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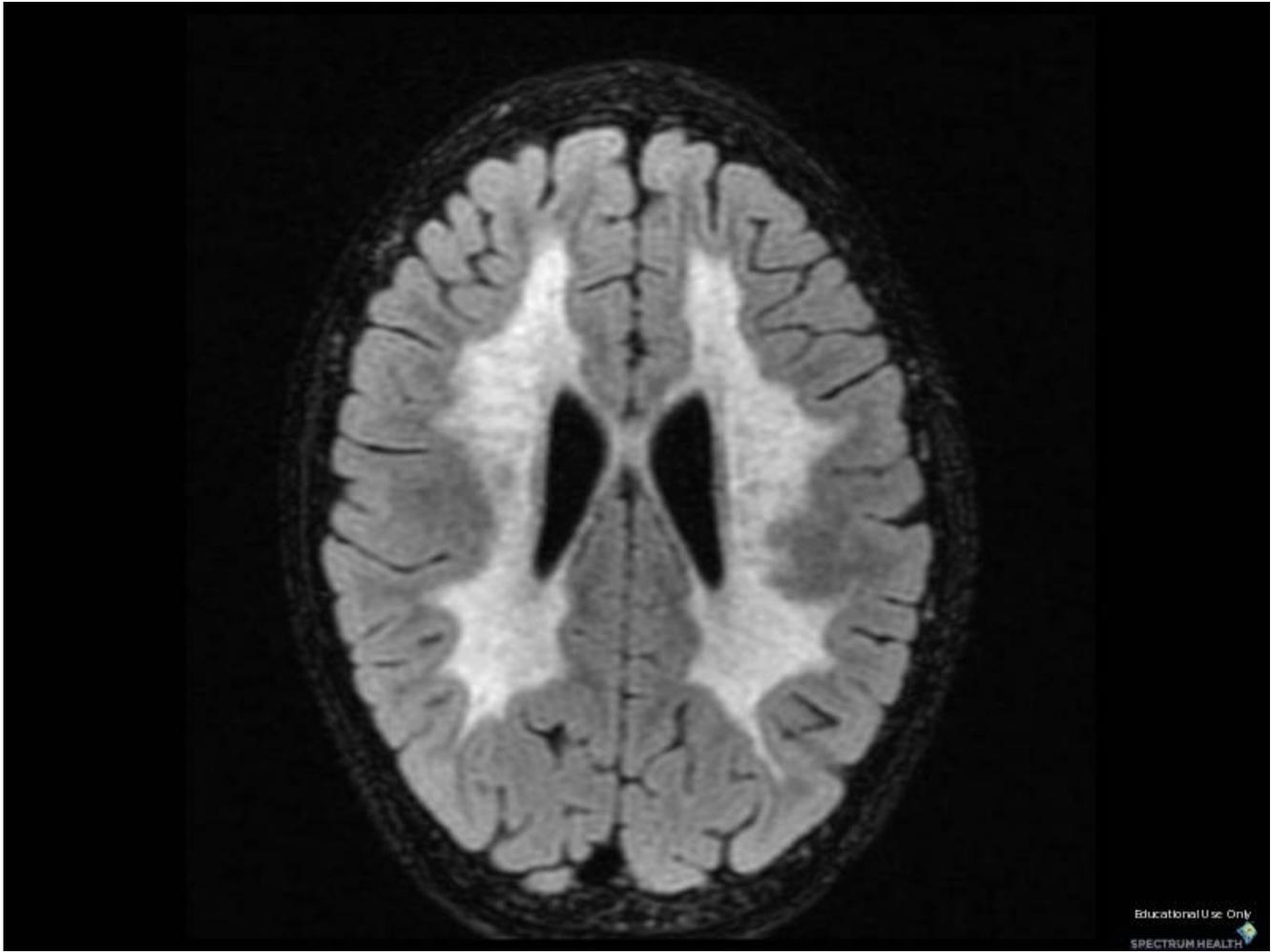












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