QUIZ 2

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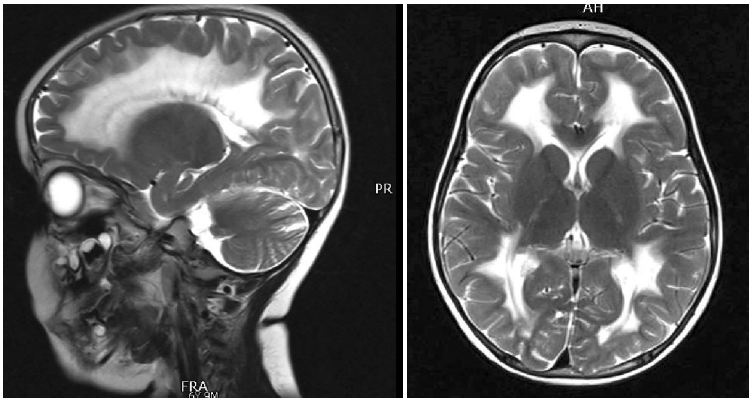
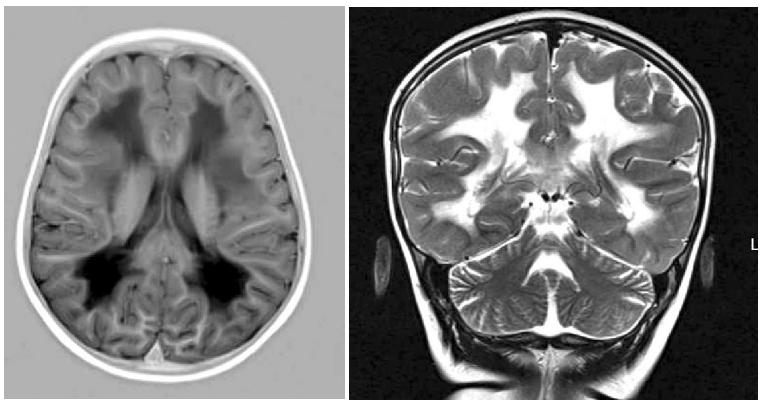
History

Child presented with motor signs of peripheral neuropathy followed by deterioration in intellect, speech, and coordination. After 2 years of on-set of the disease process, gait disturbance, quadriplegia, blindness, and decerebrate posturing was seen.





Question

Q1. What is the diagnosis?

Q2. What are the three different types of this disease and name the commonest type.

Q3. Name the characteristic dotted pattern of involvement of brain parenchyma on the images.

Q4. Which metabolite accumulates in the body tissues in this disease and how we support our radiological diagnosis biochemically?

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Answers

Answer 1: Metachromatic leukodystrophy.

Answer 2: Three different types of metachromatic leukodystrophy are recognized according to patient age at onset: late infantile, juvenile, and adult. The most common type is late infantile metachromatic leukodystrophy.

Answer 3: Tigroid pattern of demyelination. (T2-weighted MR images in these cases show linear tubular structures with low signal intensity in a radiating ("tigroid") pattern within the demyelinated deep white matter, which suggest sparing of the perivascular white matter.

Answer 4: Metachromatic leukodystrophy is an autosomal recessive disorder caused by a deficiency of the lysosomal enzyme arylsulfatase. Enzyme is necessary for the normal metabolism of sulfatides, which are important constituents of the myelin sheath. In metachromatic leukodystrophy, sulfatides accumulate in various tissues, including the brain, peripheral nerves, kidneys, liver, and gallbladder. The accumulation of sulfatides within glial cells and neurons causes the characteristic metachromatic reaction. Metachromatic leukodystrophy is diagnosed biochemically on the basis of an abnormally low level of arylsulfatase A in peripheral blood leukocytes and in urine.

References

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