ADRENOLEUKODYSTROPHY

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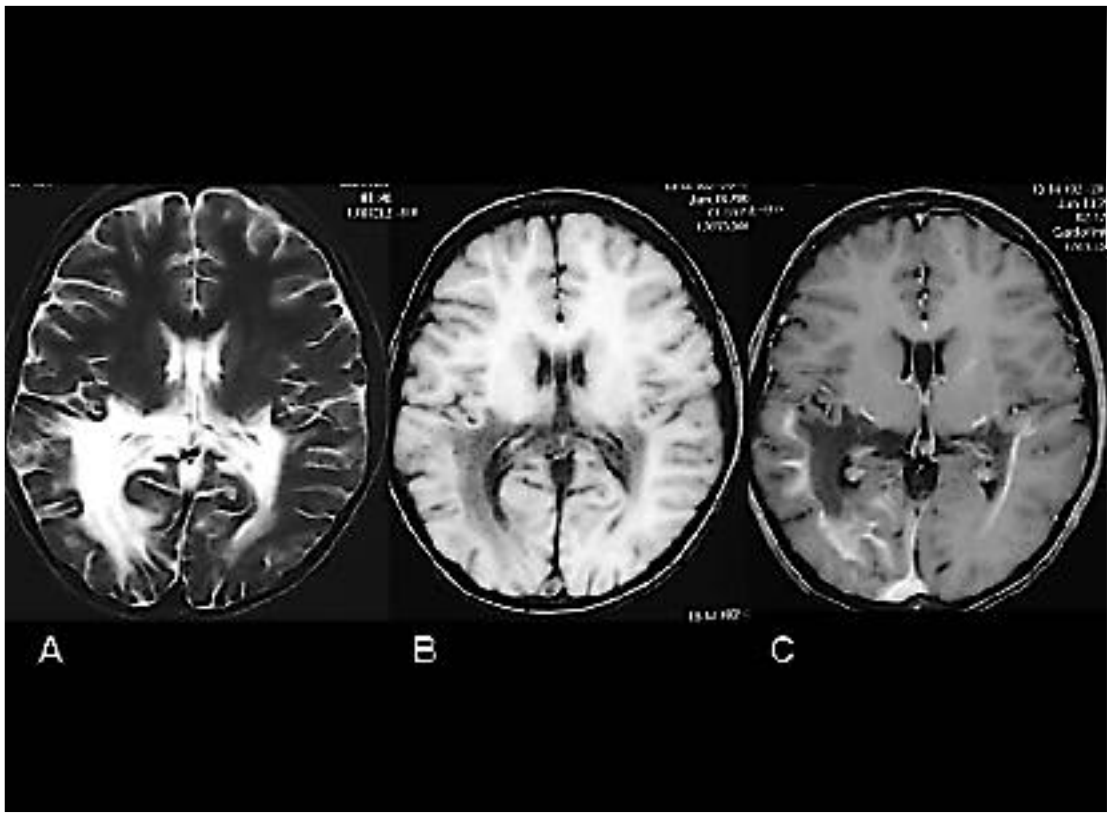

Brain MRI scans of an adolescent boy with cognitive decline. A: T2 weighted axial image at the level of the lateral ventricular trigone. There is bilateral symmetrical hyper-intense signal from the posterior white matter. The frontal white matter is completely normal. There is no mass effect. The grey matter returns normal signal. B: T1 axial unenhanced image at the same level. The affected white matter returns uniformly hypo-intense T1 signal. C: Gadolinium- enhanced T1 image shows bright enhancement at the edges of the area of abnormality. Features are typical of adrenoleukodystrophy.

DIAGNOSIS

A 12-year-old male presented with deteriorating school performance and behavioral changes over one year. His older brother had died 3 years earlier with a similar disorder. On examination, he was underweight and anemic. He was mildly ataxic and had pale discs on fundoscopy. An MR scan was requested and a diagnosis of adrenoleukodystrophy (ALD) was made.

COMMENT

Leukodystrophies are a group of disorders characterised by progressive demyelination of white matter. These are usually disorders of early childhood and typically present with regression. Most cases are fatal before the end of the first decade. ALD with its late presentation is the only dysmyelinating disorder which may be seen in young adults.

ALD is usually inherited as an X-linked recessive disorder and therefore only males develop symptoms. The defect lies in the function of linoceryl-coenzyme A leading to defective

oxidation of fatty acids and the accumulation of very long chain fatty acids in the white matter, adrenal cortex, and testes. The disease usually presents towards the end of the first decade of life with peak incidence between 8-10 years. Presentation is usually with progressive neurological and behavioral abnormalities. Vision and hearing may also be affected. The degree of endocrinological abnormalities is variable, but some adrenal insufficiency is usually present. MR scans show a progressive demyelination of cerebral white matter; the changes are bilateral, symmetric, and hyperintense on T2 weighted images. There is intense enhancement at the margins of the abnormal areas. The disease almost invariably starts in the occipital central white matter and progresses centripetally and forwards. Progression is relentless but the pace is variable. Late in the disease the entire white matter may be involved. Grey matter, however, gives normal signal even in late stages.

REFRENCES

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