Cerebral sinus venous thrombosis as the initial presentation in a child with homocystinuria: Case report

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Background: Inherited homocystinuria is a rare autosomal recessive metabolic disorder affecting several organs including the CNS resulting in neurologic and vascular abnormalities through toxic effect of homocysteine. Neurologic involvement includes progressive mental retardation, seizure, dystonia, behavioral and personality disorder, while the vascular one include stroke due to thromboembolic syndromes.

Case report: A 6-year-old boy with learning disability, skeletal abnormalities, myopia, frequent fractures and previous febrile convulsion presented with sudden onset of worsening headache, left sided frequent seizures with generalization and behavioral changes for 48 hours followed by abnormal gait and deterioration in his mental status proceeded by febrile illness. His parents were consanguineous with no neurologic disorder or epilepsy in the family. Examination revealed mental subnormality, poor social interaction, slurred speech, general hypotonia left >right, DTR+1, extensor planter response bilaterally and unsteady gait. Brain MRI and MRV showed extensive superior sagittal sinus thrombosis, localized cortical right parieto-occipital infarction and abnormal white matter of both hemispheres, sparing subcortical fibers with diffusion restriction suggestive of metabolic white matter disease. Laboratory evaluation revealed, high serum methionine, homocysteine level, and presence of cysteine in the urine. Marked improvement was noted after intravenous anticoagulant therapy. He was maintained on lamotrigine, aspirin and high dose of oral pyridoxine, folate, betaine and methionine-restricted diet. Follow-up brain MRI showed a significant recanalization of all sinuses with reduction of thrombosis within the superficial cortical veins.

Conclusion: Although homocystinuria is a vanishing disease, physician should keep high index of suspicion among patients with mental disabilities and seizure disorders as early detection and treatment can prevent future neurologic and vascular complications.

Biography

Amal Yousef Kentab has completed her MMBS from King Saud University and ABP and KSU fellowship in Pediatrics in 1996, MRCP at UK –London in 1998; Fellowship in Pediatric Neurology at King Fasial Specialist Hospital And Research Center in Riyadh in 2001. She is an Associate Professor, Consultant Pediatric Neurologist and the Deputy Director of Pediatric Residency Program at King Saud University, Riyadh, Saudi Arabia. She has published more than 35 papers in reputed journals and has been serving as a Reviewer in multiple journals and presented in multiple international conferences.

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