Metabolic Stroke; A Rare Clinical Condition of Glycerol Kinase Deficiency

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Introduction

Metabolic stroke, begins with metabolic dysfunction and leads to a rapid onset of lasting focal brain lesions in the absence of large vessel rupture or occlusion. Metabolic Stroke is routinely reported in mitochondrial diseases, glutaric, isovaleric, methylmalonic and propionic acidurias and lipid metabolism disorders, but it is a rare condition in Glycerol kinase deficiency(GKD). In this study, Metabolic Stroke, which is a rare clinical condition of GKD is presented.

Case report

7 years old boy who has GKD associated Adrenal insufficiency & Becker Type Muscular Dystrophy, developmental delay and proximal muscle weakness admitted to our hospital with alteration of consciousness. He has been taking hydrocortisone and fludrocortisone since neonatal period. He had fever and cough 3 days ago, antibiotic was started at the outpatient clinic. On the physical examination; he was unconscious(but reacts to the pain), he had neck stiffness and deep tendon reflexes were hypoactive. Because of being brain edema at cranial computed tomography, lumbar puncture couldn't performed. Acute phase reactants were high, all cultures were negative. We started ceftriaxone, vancomycin, anti-edema treatment and intravenous immunglobuline for encephalitis(infectious/inflammatory). There was brain edema, subfalcine herniation and diffuse cortical cytotoxic edema(occipital area spared) at the left hemisphere in the neuroimaging(figure 1, figure 2). Brain arteries & venouses were patent. Involvement of different blood supply areas by preserving the left occipital pole suggested metabolic infarct other than infectious etiologies. At the clinical course he had right central facial paralysis, right hemiplegia, but he started to speak and referred to the physical treatment and rehabilitation department.

Discussion

Inherited metabolic and genetic defects increase the risk of a stroke. The pathophyiology of a metabolic stroke in patients with inherited metabolic diseases are overlapping. Metabolic disorders may cause thromboembolic events other than typical ischemic stroke-associated risk factors. Additionally, in certain metabolic conditions, cellular energy failure, with consequent cell death, can occur without the cessation of blood flow. The pattern of cell death typically does not follow the classical vascular distribution because metabolic stoke is not a disorder caused by impaired blood flow; however, it may be more diffuse or random.

Metabolic stroke should be included in the differential diagnosis of any acute neurological deficits or presentation of altered consciousness levels.

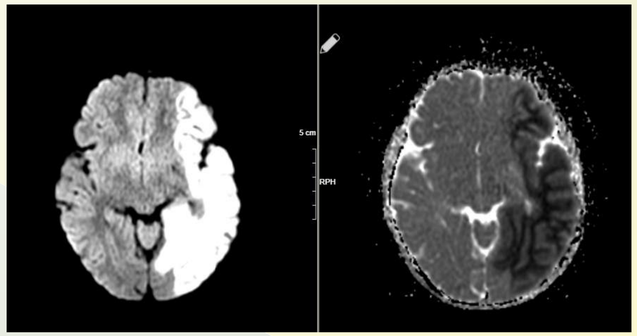

Figure 1: Diffusion and ADC images

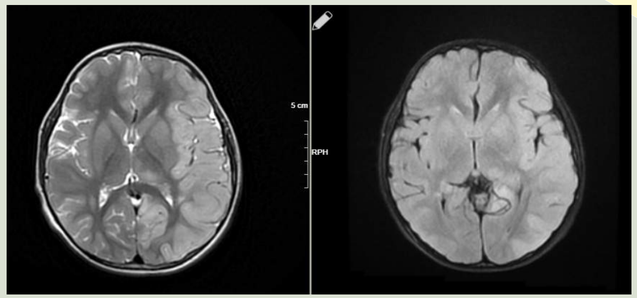

Figure 2: Axial T2 ve FLAIR images

Conclusion

At the presence of acute onset of encephalopathy and focal lesions in different blood supply areas of brain, metabolic stroke should also be kept in mind in GKD.

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