Vici syndrome: Experience at tertiary care center

Amal Yousef Kentab[1, 2]
[1]King Khalid University Hospital, Saudi Arabia
[2]King Saud University, Riyadh, Saudi Arabia

Background: Vici syndrome (OMIM: 242840) is a rare autosomal recessive congenital multisystem disorder characterized by oculocutaneous hypopigmentation, agenesis of corpus callosum, cataract, combined immunodeficiency, hypertrophic cardiomyopathy and psychomotor retardation. It is caused by mutations in the *EPG5* gene (18q12.3). Approximately 30 patients have been reported in the world. Reports of patients from the Arabian Peninsula are rare due to lack of awareness among physicians.

Method: A retrospective chart review of seven children diagnosed with Vici syndrome at a University Hospital, in Riyadh.

Results: There was female sex preponderance (71.4%). All patients were failing to thrive (85.7%) except for one. Developmental delay and hypotonia were found in all, while seizure disorder was found in 3(42.9%). Ocular albinism was detected in 2(28.6%). Facial dysmorphism was found in all (high arched palate and micrognathia) 5(71.4%) patients had optic atrophy and 4(57%) patients had retinal hypoplasia while only one had cataract. Brain MRI revealed hypoplasia of corpus callosum in 5(71.4%) patients while agenesis of corpus callosum was found in one. Hypoplasia of cerebellar hemisphere was found in all while of the vermis was found in all except one. All had delayed myelination of white matter and only 4(57%) had gyral abnormalities. Muscle biopsy was done in 3(42.9%) patients and showed myopathy. One patient had cardiac hypertrophy, 2(28.6%) patients had kidney abnormalities, and lymphopenia with lymophocytes subsets abnormalities were detected in 3(42.9%) patients.

Conclusion: Vici syndrome should be suspected in children presented with developmental delay, hypotonia and abnormalities of corpus callosum or cerebellum. Awareness may help in improving the quality of life and survival of these patients.

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.

Amkentab@hotmail.com

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