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## Isolated mitochondrial complex IV (cytochrome c oxidase) deficiency mimicking ADEM

Amal Yousef Kentab, Mohammed N Al-Nasser, Hamdy H Hassan and Hisham El Khaldi  
King Khalid University Hospital, Saudi Arabia

**Background:** Cytochrome C Oxidase deficiency is a rare inherited metabolic disorder characterized by deficiency of the enzyme cytochrome C oxidase, an essential enzyme that is active in the oxidative phosphorylation. It is clinically heterogeneous, ranging from isolated myopathy to severe multisystem presentation. Acute disseminated encephalomyelitis (ADEM) is a monophasic polysymptomatic inflammatory condition of the CNS that principally involves the white matter although the grey matter may also be affected.

**Case report:** A 6 years old boy, product of a consanguineous marriage with history of progressive weakness and convulsions noted at 2 years of age in his sister who died at the age of 5 years with no definite diagnosis. At the age of 2 years, he presented with fever, encephalopathy and seizures, preceded by varicella skin infection and diagnosed as ADEM based on MRI. He showed slow recovery but remain with poor speech and ataxic gait. At four years of age he developed the 2nd episode of ADEM, admitted in coma state and ventilated for 10 weeks. Progressive neurologic decline and frequent myoclonic seizures were noted despite full support with immunosuppressive therapy, antiviral treatment, antiepileptic drugs and megavitamins. MRI showed generalized cortical swelling and abnormal high signal intensity diffusely involving cerebral cortex, cerebellum, and deeper gray matter. He remained in vegetative state with spastic quadriplegia. Muscle biopsy showed reduction of cytochrome C oxidase activity and abnormal accumulation of mitochondria at subsarcolemmal membrane.

**Conclusion:** This case serves as a reminder of the clinical heterogeneity of cytochrome C oxidase deficiency, and physician should consider a broader differential diagnosis for ADEM.

### Biography

Amal Yousef Kentab has completed her MMBS in 1989 from King Saud University; ABP and KSU fellowship in Pediatrics in 1996, MRCP at UK - London in 1998; fellowship in Pediatric Neurology at King Faisal 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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