Diaphragmatic Myoclonus

We diagnosed abdominal myoclonus in an 8-year-old boy with terminal stage of neuronal ceroid lipofuscinosis (NCL). He was normal up to two years of age when he had onset of myoclonic seizures. Over time his seizures worsened. He lost speech and mobility by six years. Recently he was admitted with recurrent seizures and frequent jerking of the abdominal muscles. His three siblings had died of similar illness. Examination revealed choreathetoid movements of limbs, multifocal myoclonus, and diaphragmatic myoclonus (***Web Video* I**). Surface electromyography (EMG) electrodes applied on the abdomen wall revealed around 120 contractions per minute. Electroencephalography (EEG) showed bihemispherical periodic discharges at rate of 1.5 to 2 per second, left side more than right. Intravenous bolus of phenytoin followed by midazolam infusion controlled his abdominal (diaphragmatic) contractions.

Diaphragmatic myoclonus (flutter) is an abnormal movement of diaphragm resulting in inward and outward movements of abdominal wall synchronous with diaphragm contractions. In case other respiratory muscles are involved, it is called respiratory myoclonus. Other names like moving umbilicus syndrome, and belly dancer's dyskinesia have also been given. The diagnosis of diaphragmatic movements can be confirmed by EMG of diaphragm, or on fluoroscopy. The origin of the myoclonus is believed to be the brainstem, basal ganglia or cortex.

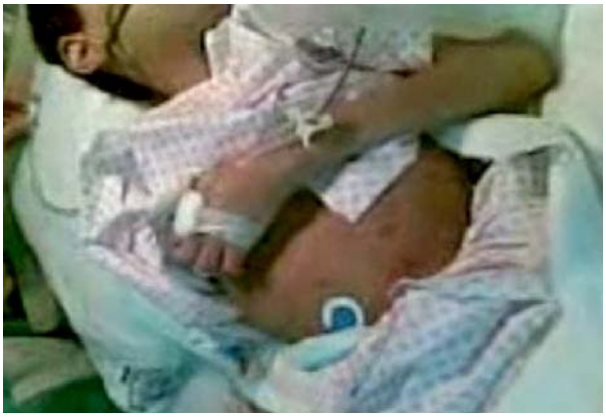

FIG. 1 *Diaphragmatic myoclonus in a child (see Video at website).*

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