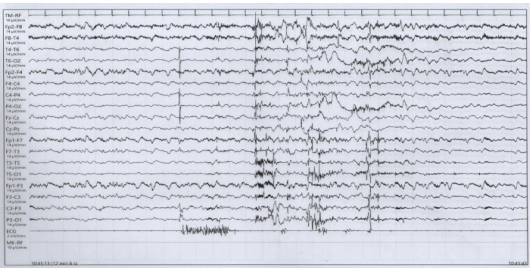
SEVERE HYPOTHYROID ENCEPHALOPATHY IN A PATIENT WITH DOWN SYNDROME: IT IS NOT ALWAYS LOMEDS

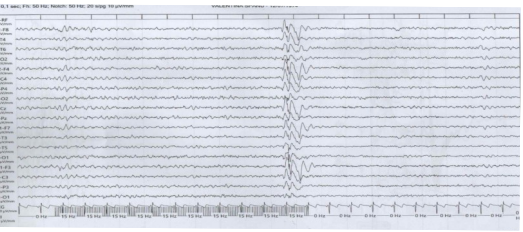
Autori: S.Collu, F. Orlandini, G.Serra, D.Murgia, G. Cossu
Università degli Studi di Cagliari, ARNAS Brotzu (CA)

Introduction: In patients with Down Syndrome, epilepsy is frequently observed and has a prevalence of about 46% in patients over 50 years of age, which is higher than in the general population (1-13%), but lower than what is found in individuals with intellectual disability. In patients with DS, epilepsy has a bimodal onset (childhood and after age 50 due to cerebral structural alterations comparable to those found in patients with Alzheimer).

Case: A 47-year-old woman with DS, with no prior history of epilepsy, was admitted for GTCS (Generalized Tonic-Clonic Seizure).She presented with severe cognitive decline, congenital strabismus, and recent surgery for evacuation of a post-traumatic SAH (Subarachnoid Hemorrhage). Upon admission, she was in a comatose state (somnolent, motor responses to pain, normo-reactive pupils). Brain CT showed cortical atrophy and ventriculomegaly. The EEG showed diffuse slowing in the absence of clear epileptic abnormalities. The metabolic profile revealed severe hypothyroidism (TSH 87.22); in light of these data and after excluding other possible causes, a diagnosis of myxedema coma was made. After administration of Levothyroxine, her consciousness rapidly improved. The residual myoclonus was managed with antiepileptic therapy. Subsequently, the patient developed a septic condition with the consequent onset of sub-entrant epileptic seizures leading to status epilepticus, which were poorly responsive to antiepileptic therapy and resulted in her death a few days later.



Above patient's EEG ; Below LOMEDS's EEG



	HYPOTHYROIDISM	LOMEDS
ONSET	Acute/ sub- acute	Late onset
SYMPTOMS	Respiratory depression, loss of consciousness, hypo/areflexia, convulsions, cognitive decline	Myoclonus upon waking, sometimes generalized seizures.
COURSE	variable	chronic
PROGNOSIS	treatable/reversible	Death within a few years of diagnosis
EEG	slowed and altered cerebral activity, with the presence of slow waves (<5 Hz) and spike-wave discharges, typically generalized and bilateral	background activity that progressively slows down and a prevalence of paroxysmal activity in the temporo-frontal regions, with frequent presence of generalized and bilateral spike-polyspike-wave complexes

Conclusion: In patients with Down Syndrome and epileptic seizures, there is a tendency to promptly diagnose LOMEDS, a form of epilepsy with a chronic course, associated with cognitive decline and myoclonic seizures/GTCS. However, an acute neurological worsening requires the exclusion of reversible conditions, such as metabolic disorders

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