

Azienda Ospedaliero-Universitaria "A. Meyer"
UOC Neurologia Pediatrica - Università degli Studi di Firenze
Direttore Prof. Renzo Guerrini
Viale Pieraccini 24, 50139 Firenze
Tel. +39 055 5662835/2834

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Clinical Report

De Santis Maurizio
D.o.B: 05/03/2018

Maurizio is a 2 year old child affected by a neonatal epileptic encephalopathy with pharmaco-resistant focal seizures and spasms. The clinical picture appeared to be caused by a *de novo* mutation c.1936A>G in the *GRIN2A* gene. The mutation is thought to be *gain of function*. The brain MRI performed at the age of 4 months was normal.

The patient has had a 18 month follow-up at our hospital.

Maurizio shows a severe psychomotor delay. PEG carrier since October 2019. At the neurological examination, eye contact is brief and inconstant. Bilateral esotropia. Oriented by sound stimulation. No verbal language ability. Muscular hypotrophy, axial hypotonia. He cannot sit alone. Dyskinetic motor pathway.

The child presents daily focal seizures, tonic seizures and spasms, both in brief multiple clusters and isolated, mostly concentrated during sleep and upon awakening. Currently the child assumes Memantine at the dosage of 0.8 mg/kg, Carbamazepine, Phenytoin and Cannabis flos 9% CBD, 0.4% THC. The child has been following ketogenic diet since June 2020.

We have performed many prolonged EEG recordings, showing a slow disorganized background activity with many multifocal sharp-wave abnormalities during sleep, both synchronous and asynchronous.

Seizures have been treated with many anti-epileptic drugs, both alone and in combination with very low or transitory therapeutic effects (Carbamazepine, Levetiracetam, Pyridoxin, Clobazam, Clonazepam, Nitrazepam, Phenobarbital, ACTH, Hydrocortisone, Vigabatrin, Valproic Acid, Betametasone ev, Topiramate, Rufinamide, Perampnel and Memantine). We have observed a clinical benefit from treatment with Memantine up to the dosage of 0.6 mg/kg, which has been started in monotherapy in November 2018 until March 2019. During this period, focal seizures have progressively reduced their frequency. Between January and March 2019, no focal seizures have been reported by the family. Spasms have always been appreciated daily.

A recent capnography has resulted normal.

Dr. Martino Montomoli, MD
Dr. Norman Panza, Resident

