

CASE STUDY 6: H.M.A.M

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Patient Profile

H.M.A.M., DoB 19.10.2021, Male

First born of healthy first cousin parents, normal pregnancy and birth with delivery at term. Neonatal check-up on day 4: feeding disorder, incidental episodes to be defined. At the anamnesis, 2 siblings of the mother died at 2 days and 2 months of age from unknown causes.

Background

He was admitted to neonatal pathology and a neurological physical examination reported an interaction not assessable, fixation absent. Head free, CC 33.5 cm (16thpc.le). Nc uninjured. Hypertone to the 4 limbs and axis, poor spontaneous motility, chaotic, jumpy. Neurological objectivity included apostural tetraparesis, microcephaly, visual deficit of central origin.

Clinical Assessment

From the video EEG recording and polygraphy, the child presented intercritical pattern: asymmetrical and asynchronous suppression-burst (SB).

Clusters of rapidly occurring seizures reported diffuse tonic component, axis torsion, eyeball version, asynchronous shaking clonias and critical pattern has polymorphous multifocal and bascular. Encephalon MRI and Spectroscopy was within limits with no neurometabolite peak abnormalities.

At 12 months the skin biopsy was negative and the muscle biopsy showed reduced activity of Complex I of Respiratory Chain Complex I: NADH CoQ1 Rd at 10.6 (reference values 17.0-34.0). Therefore, the definitive diagnosis remains open, and it is treated for the early onset epileptic encephalopathy. Search for a rare disease or a rare phenotype or a new gene is still open using the tools available today: ArrayCGH, Whole Exome Sequencing, Whole Genome Sequencing and, above all, time.

Dietetic Intervention

Therapy consists in trial with Vitamin B6, pyridoxalphosphate, folinic acid, Phenobarbital EV + Phenytoin EV.

The Ketogenic Diet (KD) 3:1 started since the age of 2-months with an infant formula 1 mixed with a ketogenic formula 4:1 and water, calorically adjusted according to the child's growth and increased nutrients' needs. The diet was supplemented with vitamins, minerals and combined with potassium citrate.

At 7 months a 3:1 KD of 650 kcal/day was planned for weaning with 7 daily meals about every 3 ½ hours, with creamy meals made from fresh and homogenised food mixes, and it was emphasised to maintain adequate hydration according to the dietary prescription.

Clinical Outcome

Throughout the diet period, the child had no seizures, the dietary compliance was optimal and meals were consumed without any difficulty and no gastrointestinal symptoms. Ketonemia was stable around 1.5-2 mM/l and no alterations in blood glucose have been found. Nutritional status was characterized by adequate weight and body growth in accordance with percentiles of the general population.

A slight increase in uric acid (7.4 mg/dl) occurred at 6 months but it receded at subsequent follow-up as did the hint of cholelithiasis investigated by ultrasonography. Lipid profile was always in the normal range.

Learning Points

The family reported good tolerance and acceptance and feels motivated by the absence of crisis and the maintained optimal growth of the child.

