

EXAMINATION REPORT

NAME: MAIA ANGHELINA

PERS. ID NO.: 6180427410150

DATE OF BIRTH: 27-Apr-2018

DATE OF EXAMINATION: 16-Apr-2019

REFERRING PHYSICIAN:

EXAMINATION: Paediatric neurology examination by Dr. Barca

FINDINGS:

Maia, a female infant aged 11 months and 2 weeks, presented for neurological examination.

- The girl was born after a risk pregnancy (bleeding, short cervix), from mother diagnosed with thrombophilia from the GA of 27-28 weeks, with initiation of Clexane therapy, with bed rest during the last weeks of pregnancy, birth at the GA of 39 weeks by C-section, W 3,270 g, APGAR score 9/10, good postnatal adaptation, discharged from the maternity hospital after 3 days.

- At the age of 3 and ½ months, parents were alarmed by the occurrence of short episodes described as a “tremor” of the extremities, noticed during meals

- The EEG showed right focal epileptic seizures

- The parents asked for a second opinion in Turkey – where epileptiform discharges were identified as corresponding to the clinical manifestations

Therapy with Levetiracetam 450 mg daily was initiated; from 300 mg daily, no more seizures were observed (from November 2018)

- Despite the seizure control, the EEG appearance worsened – hypsarrhythmia was noted, for which VGB 750 mg daily was introduced in the therapy regimen (the dose could not be increased because of the somnolence effect, which is currently decreasing)

- Currently, Synachten was proposed – the parents have not reported any more crises, but altered EEG traces were observed

The patient did not show any obvious regression, but the parents describe “good” and “less good” days (when the girl is less alert, showing increased movement disorder)

- Comprehensive investigations:

- Normal brain MRI scan (performed at the age of 4 months and 3 weeks)

- Laboratory tests showed a normal blood glucose/CSF glucose ratio

- Serum aminoacids, urinary organic acids

- Extended neonatal metabolic screening – normal

- Family history: maternal grandmother’s brother had 2 “seizure” episodes

Cranial circumference 44 cm, W 9.2 kg, harmoniously developed body.

Active infant, showing good mood, however she does not react when calling her name and makes suboptimal eye contact. Slightly jerky eye movements, however when she succeeds to fix the target, she follows all axes. Slight sialorrhea.

- At traction in the sitting position the head is on the trunk axis, axial titubation when sitting, short startles causing the head to fall (myoclonus of the type negative myoclonus? trunk ataxia?); during the examination, short movements of the eyeballs (oculogyric crises?); rolls, rotates around her longitudinal axis; sits when seated in the sitting position, but does not move; parachute reflex +, symmetrical; can hold 2 toys at the same time, banging one against the other;

- Makes gurgling sounds, does not babble.

Developmental age: 6-7 months.

PERSISTENT EEG CHANGES: APPEARANCE OF EPILEPTIC ENCEPHALOPATHY. MILD GLOBAL DEVELOPMENT DISORDER, WITH ATAXIA ELEMENTS OF UNKNOWN AETIOLOGY.

Recommendations:

1. Continue kinetotherapy.
2. Continue Levetiracetam 4.5 mL daily and follow the Synachten therapy regimen with the initial dose of $\frac{1}{2}$ from a 1 mL/mg vial) recommended by the neurologist who monitors the baby (Dr. Tarța-Arsene).
3. Filming of paroxysmal manifestations.
4. Perform genetic testing of the type whole exom sequencing (WES)

Dr. Diana Barca
Paediatric Neurologist
987895

[physician's stamp]
Asist. Univ. DIANA BÂRCĂ
Senior Physician, Paediatric Neurology
987895
[illegible signature]

[Medical unit's stamp]
SOCIETATEA COMERCIALĂ
HIPERDIA S.A.
12
BRAȘOV – ROMANIA

The undersigned, GOIA ALEXANDRU, certified translator under no. 23764/2008, hereby certify the accuracy of the translation into English of the photocopy of authentic document written in Romanian, which has been seen by me.

