SLEP SUMMER SCHOOL 2020
Fellow 8 Thais Milioni Luciano

Patient first visit at 13 days old, for neonatal screening evaluation (TSH=10.4 mUI/ml). In the occasion the mother referred that the child was breastfeeding exclusively, with good suction, and not yet recovered her birth weight (he was 120g down the birthweight). She had no other complaints.

Background:
Birth by cesarean, GA: 39w, weight 3740 g, length 47 cm, Apgar 9/10, no birth complications. Mother denies any complications during pregnancy.

Family background:
Mother (32 y), father (41 y), brothers: 11 y (only mother's son) and 14 y (only father's son), healthy. Brother, 18 y (only father's son): epilepsy.

Physical exam:
No alterations at physical exam. No neck malformations, thyroid not palpable. Typical male genital, scrotal testicles. Primitive reflexes present and symmetrical.

Confirmatory tests:
TSH: 22.8 mUI/ML, total T4: 6.3 mUI/ml
Thyroid scintigraphy (technetium): eutopic thyroid.

Diagnosis: congenital hypothyroidism

Management: levothyroxine (6.9 mcg/kg/day). The thyroid function normalized and was adjusted at each return as needed.
At 2 years old: patient developed obesity, which was associated with a large intake of calories.
At 2 years and 6 months: delay in language and cognitive development.
At 3 years old: patient presented seizures and cramps. He started evaluation with pediatric-neurology group, which confirmed the cognitive and language development delay and initiated anticonvulsant therapy, with only partial control of seizures. An electroencephalogram was performed with no characteristic findings of specific diseases. By the association of hypothyroidism, developmental delay and partially controlled seizures, an exome was request.
At 4 years: during the endocrinology follow-up, mother complained of persistent cramps and poor control of seizures