

Use of topiramate in severe hyperphagia associated to neuropsychiatric features in a boy with congenital proopiomelanocortin deficiency (POMC)

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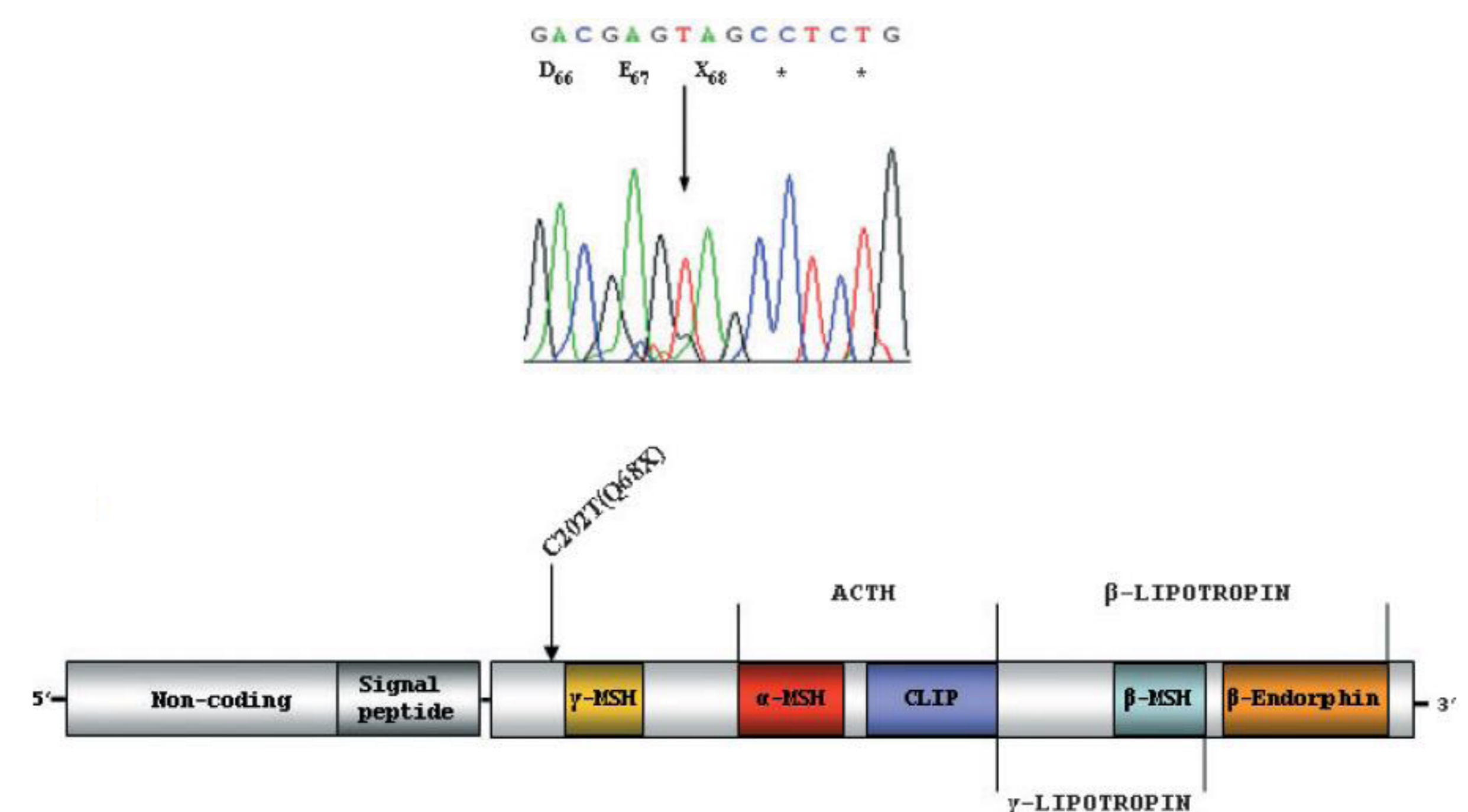


Disclosure statement: nothing to disclose

POMC gene mutations cause early-onset obesity, hyperphagia and ACTH deficiency. In subjects with this picture, neuropsychiatric (NP) features were rarely reported.

An 8-month-old Egyptian male infant was referred for severe early-onset obesity due to severe hyperphagia. Isolated ACTH deficiency was detected and treated with Hydrocortison since he was 10-month-old; cerebral and hypothalamic-pituitary MRI were normal.

Direct sequencing of the POMC gene revealed a homozygous single substitution C6902T determining a Gln68X substitution. Therefore congenital POMC deficiency was diagnosed.

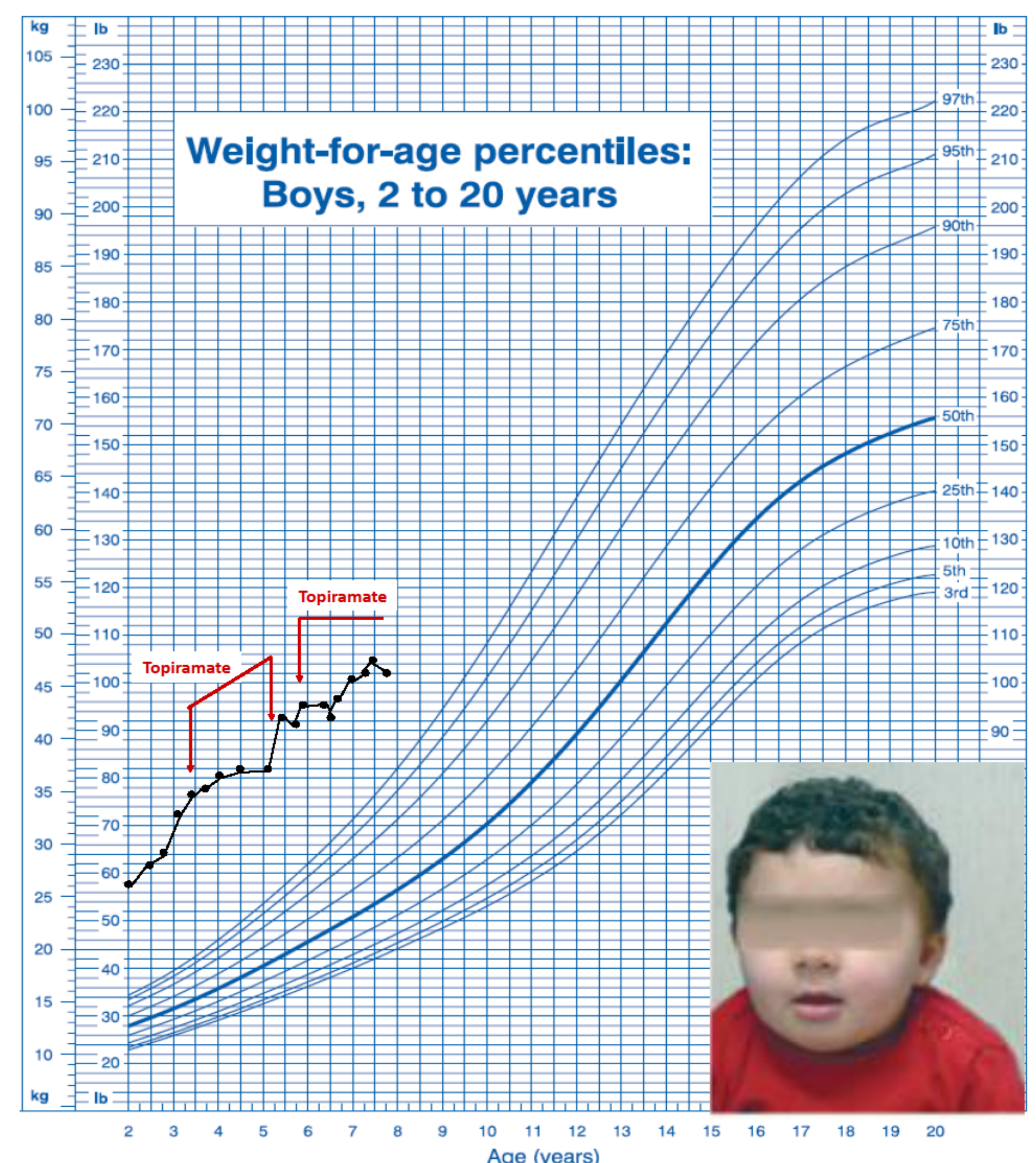


The child spoke his first words and walked alone at 2 yrs of age. Since 2.5 yrs, he was followed for neurocognitive development and psychiatric aspects. At the first evaluation, a neurodevelopmental delay was noticed by the Griffiths Mental Development Scale (Developmental Quotient 64).

Many Authors described an appetite loss during the topiramate treatment. When the boy was 3.5-year-old, due to the worsening hyperphagia associated with an oppositional defiant disorder, a treatment with topiramate was started with a good response on hyperphagia and behaviour disorder; no side effects were reported.

At 6 yrs, an attempt to stop topiramate caused a rapid worsening of hyperphagia and NP symptoms leading to a resumption of the therapy. During topiramate therapy, BMI has not increased.

At last NP evaluation at 6.5 yrs, WPPSY-III cognitive test evidenced a borderline total IQ (87) and significant discrepancy between verbal and performance IQ (verbal IQ: 70, performance IQ: 111). Teacher and Parent Questionnaires (CBCL and CTRS-R:L) revealed attention disorder and confirmed the oppositional defiant conduct.



In patients with congenital POMC deficiency hyperphagia can contribute or be associated to NP disorders. Topiramate therapy could be considered.

