Overweight: The Clue to Pseudohypoparathyroidism

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A 4-year-old girl was referred to a central pediatric hospital with overweight history. She had a family history of mother with hypothyroidism without neurodevelopmental disorders registered and past medical history of preterm birth (gestational age 34 weeks), light for gestational age (birth weight 1610 g, percentile 10 of Fenton preterm growth chart), recurrent wheezing, attention deficit disorder associated with developmental coordination disorder, specific language impairment and speech sound disorder. Her weight has evolved on the percentile 50 until 3-year-old and then increased to the percentile 75-90. Height showed progressive recovery from the first year of age and remained stable on the percentile 25 since 4-year-old (Tanner-Whitehouse curves).

On the first appointment, physical examination revealed weight 18.4 kg (percentile 90), height 98.5 cm (percentile 25), body mass index 18.9 kg/m^2 (percentile 85-90), trunk adiposity, rounded face, enamel hypoplasia, small hands and apparent shortening of the fourth and fifth metacarpals, bilaterally (Fig. 1). Similar hand findings were found on her mother (Fig. 2).

Laboratory investigation revealed low free-thyroxin levels (5.1 ng/dL; reference values 5.98-14.7 ng/dL), elevated thyroid-stimulating hormone (14.6 µg/dL; reference values 0.7-5.97 µg/dL) and increased parathyroid hormone levels (289 pg/mL; reference values 14-72 pg/mL). Serum phosphorous and calcium were normal (respectively 5.8 and 9.4 mg/dL).

We diagnosed our patient with pseudohypoparathyroidism and she started therapy with calcitriol and levothyroxine. She was referred to perform genetic study, which awaits.

In most cases, pediatric overweight is caused by a sedentary lifestyle and eating behaviors. However, it may be related to endocrine pathology, genetic syndromes or drugs, among other rare causes.[1]

Pseudohypoparathyroidism is a heterogeneous group of rare endocrine disorders characterized by resistance to the action of parathyroid hormone. There are five variants of pseudohypoparathyroidism and most has identified genetic abnormalities.[2] Pseudohypoparathyroidism type 1a is the most common subtype and is associated with phenotypic characteristics, including overweight, short stature, rounded face, shortened metacarpals and other bones of the hands and feet.[2-4] A loss of expression of the intracellular messenger, common with thyroid-stimulating hormone action, justify the existence of primary hypothyroidism, as we observed in our case.[3] Genetic testing can confirm diagnosis and identify subtype.

This clinical case illustrates a rare endocrinological cause of overweight in pediatric age, whose early diagnosis avoids the consequences of persistent or intermittent hypocalcemia[5]. Clinical suspicion with recognition of phenotypic aspects is essential.

Keywords: Brachydactyly; Child; Overweight; Pediatric Obesity/etiology; Pseudohypoparathyroidism/complications; Pseudohypoparathyroidism/diagnosis

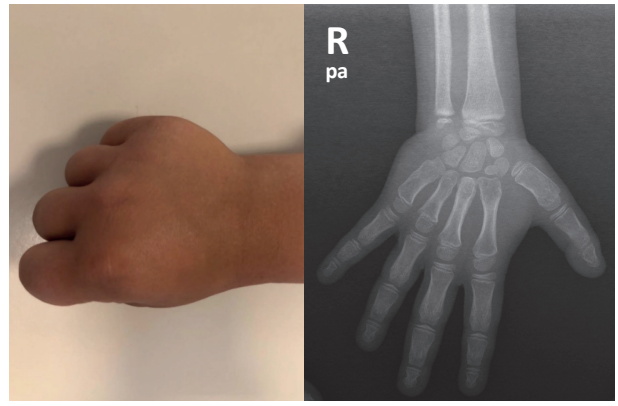

Figure 1. Brachymetacarpia of the fourth and fifth metacarpals, confirmed by hand radiography.

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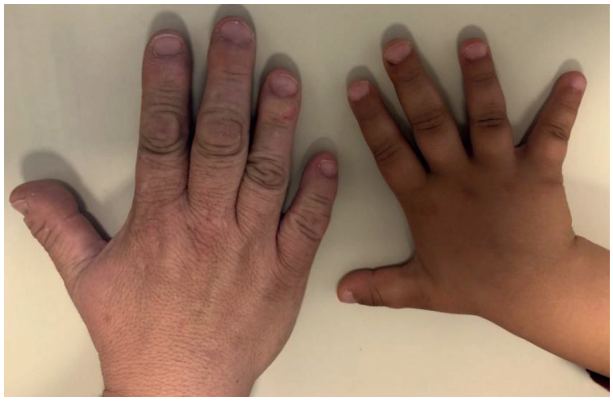

Figure 2. Comparison of metacarpal changes (fourth and fifth) in the hands of the child and her mother.

Conflicts of Interest

The authors declare that there were no conflicts of interest in conducting this work.

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Confidentiality of data

The authors declare that they have followed the protocols of their work centre on the publication of patient data.

WHAT THIS REPORT ADDS

- Overweight and obesity in pediatric age, although infrequent, may be associated with genetic or endocrine causes.
- Specific phenotypic characteristics should raise the suspicion of underlying pathology, so a detailed physical examination is essential.
- Pseudohypoparathyroidism is a rare endocrine disorder, characterized by resistance to the action of parathyroid hormone, with implications on phosphorous and calcium metabolism.
- Early diagnosis is essential for timely treatment and minimization of potential impairment of bone health.

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