TRICHO-RHINO-PHALANGEAL SYNDROME TYPE 1 IN A GIRL WITH GROWTH HORMONE DEFICENCY

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BACKGROUND

Tricho-rhino-phalangeal syndrome Type I (TRPS I) is a rare autosomal dominant genetic disorder clinically characterized by craniofacial and skeletal abnormalities, associated with cone-shaped epiphyses, brachydactyly and short stature.

Although patients with TRPS I present various degrees of short stature, there are only four reports of growth hormone deficiency in patients with TRPS I.

We present the case of TRPS I and partial GH deficiency

CASE REPORT

A 15-year-old female was referred to our clinic for short stature. Her height was: 141 cm (-3.7 SD), and weight: 46 kg (-1.1 SD).

Physical examination showed sparse hair, protruding ears, bulbose nose with long filtrum, significant shortening of bilateral metacarpal and metatarsal bones. Neuromotor development was normal. She had menarche when she was 11 years old.

Biochemial and metabolic test results were normal. Thyroid functions were normal as well. Bone age was 15 y according to Greulich and Pyle method.

X Rays revealed brachydactyly, cone-shaped epiphyses, Perthes-like changes of the femoral head, coxa plana and coxa magna. DEXA scan corrected for height age showed Lomber spine Z score of -2,9.

Growth hormone (GH) stimulation tests showed low response after clonidine (GH peak of 6.2 ng/ml) and levo-dopamine (GH peak of 4.1 ng/ml) administration.

Sellar MRI showed a normal size and location of the pituitary gland and stalk.

CONCLUSION

TRPS I is a rare genetic disorder characterised by typical craniofacial and skeletal abnormalities.

Short stature is a common characteristic of TRPS patients.

This is the fifth report of a patient with TRPS I showing partial GH deficiency.

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Syndromes : Mechanisms and Management

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