BACKGROUND AND OBJECTIVES

**Pseudohypoparathyroidism (PHP)** refers to a heterogeneous group of rare endocrine disorders caused by genetic or epigenetic abnormalities affecting the GNAS locus. It is mainly characterized by resistance to PTH and TSH and a complete or partial Albright Hereditary Osteodystrophy (AHO) phenotype. Few data so far exist on GHRH and LH/FSH resistance, calcitonin resistance and glucose-lipid metabolism involvement, as well as on neurocognitive aspects. The recently published healthcare-pathway proposed by the Italian Society of Pediatric Endocrinology and Diabetology (ISPED) has standardized the clinical approach to these patients. The present aim is to assess the adherence to the healthcare-pathway of the main Italian Pediatric Endocrine Centres, outlining the current state of follow-up and to obtain preliminary data on prevalence, correlation and evolution of PHP features, above all the less studied ones.

RESULTS

The average age at diagnosis in patients with mutated GNAS gene is lower than in patients with WT gene (5.18 vs 9.25 y.o., p=0.049). Pregnancy and/or delivery complications and SGA were reported in 68 and 18%, respectively; overweight or obesity (average BMI 23.32, +1.44SDS) and height <-2SDS (average 139.8 cm, -1.12SDS) in 61% and 24%, respectively. In 4/23 patients the GH-stimulation test has confirmed a resistance eligible for rHGH-therapy, whereas in 35% IGFI was <5SDS. Some age was advanced in 69% of cases.

**Focus on less studied PHP features**

A FSH/LH resistance has been found in 31% of the whole cohort and calcitonin increased levels in 55% of patients, all with mutated gene. Glucose metabolism was normal in 95% of patients, while 22% showed hypercholesterolemia, 11% hypertriglyceridemia and 78% HDL-C reduced levels. Finally, subcutaneous ossifications have been described in 32% of patients, delayed neurocognitive development in 63%, psychological support needs in 22%.

CONCLUSIONS

ISPED healthcare-pathway hasn’t been completely adopted yet, but it is confirmed to be a valid instrument to conform the clinical approach to PHP patients, with the dual aim to guarantee the best management to all patients and to allow homogeneous data collection, useful for a disease with heterogeneous and complex clinical manifestations, such as Pseudohypoparathyroidism.