Conclusion

➢ The consensus statement covers recommendations for the clinical and molecular diagnosis, as well as the management of patients with pseudohypoparathyroidism (PHP) and related disorders.

➢ A coordinated and multidisciplinary approach from infancy through adulthood should help us to improve the care of patients affected by these rare disorders.

Methods

Thirty-seven participants from thirteen countries were invited to participate in the development of this consensus statement, based on their publication results and international accepted expertise. Experts included representatives from six international societies (ECTS, APPES, ESHG, PES, ESE and ESPE), two European reference networks (ENDO-ERN, BOND-ERN) and a European network on imprinting disorders (COST BM208); further, from patient support groups (AEPHP, K20, ACRODYS group, IPOHA).

A comprehensive literature search was conducted including articles published from January 1990 through December 2016. We reviewed over 800 articles in three working groups (clinical diagnosis, molecular diagnosis and management). The preparation for the consensus took over 24 months and included two pre-meetings.

Voting: A: evidence or general agreement allows full agreement to the recommendation, B is in favor, C is weak, D: there is not enough evidence or general agreement to agree with the recommendation. If the majority was D, the recommendation was not accepted. Proportion of votes received by the option with the most votes, the strength of the recommendations was recorded as follows: +26-49% of votes, +50-69, +70+. The representatives of the patient support groups took part on the discussions, but not on the voting.

Working group 3 (Management of patients)

Patients should be screened at diagnosis and during follow-up for specific features, such as PTH resistance, TSH resistance, growth hormone deficiency, hypogonadism, skeletal deformities, oral health, weight gain, glucose intolerance of type 2 diabetes and hypertension, as well as subcutaneous and/or deeper ectopic ossifications and neurocognitive impairment. See ref. 2 (Nature reviews Endocrinology 2018) for details.

* We developed 14 recommendations in WG1, 13 in WG2 and 40 in WG3. Here we present only a very small part of our results.

Reference

- From pseudohypoparathyroidism to inactivating PTH/PTHrP signalling disorders (PAPPSD), a novel classification proposed by the EuroPHP network. Euro J Endocrinol 2016 Dec; 175(6): 1-17