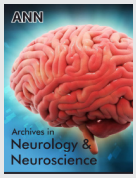

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Research Article

Pseudo Hypoparathyroidism Narrative Revue

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Abstract

The term pseudohypoparathyroidism was first introduced in 1942 by Albright to describe patients who presented with hormonal parathyroid disease (PTHb-resistant hypocalcemia and hyperphosphatemia) with characteristic skeletal and developmental changes. Pseudohypoparathyroidism is a group of rare, related and highly heterogeneous disorders characterized by target organ resistance to the action of parathyroid hormone. PHP and related disorders are caused by genetic and/or epigenetic changes leading to downregulation of a cyclic adenosine mono phosphate generator, primarily linked to the Nucleotide binding protein, Alpha Stimulating activity gene. As a story on the disease; It is under the name of a curious eponym referring to an old breed of dwarf chickens created by Sir John Sebright, in the 19th century, that the American endocrinologist Fuller Albright (1900-1969) described for the first noted in 1942 the " Sea brightbantam syndrome", a morphological syndrome characterized by short stature, obesity, rounded facial features, finger length abnormalities, often accompanied by mental retardation, associated with hypocalcemia and hyperphosphatemia.

A criteria-based narrative systematic literature review PRISMA-P has summer carried out in leaning methodologically on there publication of the article of Zaugg And al as well as the guide to recommendations for journals systematic narrative of the Economic and Social Research Council. There review of there literature has summer performed In THE basics of data international MEDLINE and Cochrane data base of systematic reviews by their search engine (respectively Pub Med). The data French speakers were searched with the search engines of the Database of the Scientific Literature in Health site (LiSSa), of the University Documentation System (SUDoc), the Catalog and Index sites Medical of language French (CISMeF). Selection of the double studies _ blind _ results products by the app of the equations of research In the different search engines made it possible to obtain a list of publication. The selection was initially made on the title and the abstract and In A second time on there reading of Full Text. As Criteria of inclusion And of exclusion studies were to be written in language English or French. Criteria major of inclusion were to be found In THE title or THE summary: Subject treating pseudo hypoparathyroidism. The exclusion criteria for the first selection phase based on title and summary were non-response to the inclusion criteria and the absence of summary available. The extraction of data After pooling publications included or excluded by title and summary, THE studies have summer read in their entirety of manner independent. Data was extracted using a data extraction grid developed from the Cochrane and PRISMA-P repositories. This second stage has permit there selection definitive of the items Following the same methodology as in the first step. We have could formalize THE points analyzes following: Title, Author Journal, Population of the study (kind actor, country, number of topics) ; the year of the study (or by default, year of publication); Objective main of the study as well as the Methodology used. The analysis of there quality of studies: The level of quality of each study was assessed using different scales: COREQ guidelines for qualitative research, criteria STROBE for descriptive studies, the ENTREQ criteria for qualitative research syntheses and the PRISMA-P criteria for THE journals of literature. description _ of the studies selected with search equations and key words used in the different bases data made it possible to reference 239 publications. After analysis 113 documents have been retained by consensus. The elements retained were distributed as follows: 55 articles and 58 academic documents (thesis or memory) obtained from the research equations. As results found:

In Japan, a national epidemiological study in 1998 estimated that there were 430 (95% confidence interval 330–520) patients with PPH in Japan, equivalent to a prevalence of 0·34/100,000.The study showed that a majority of women (58%), and the majority of patients were diagnosed during the first 4 decades of life.

In Pathophysiology, indirect evidence makes it possible to eliminate a defect in renal alpha-hydroxylase as a determining factor in this condition. Clinical signs: signs of hereditary Albright osteodystrophy, progressive bone heteroplasia, acrodysostosis, cognitive disorders Intracerebral calcification. PTH resistance should be treated with activated forms of vitamin D, for example calcitriol or alfacalcidol, to increase serum calcium levels and thereby reduce PTH levels. The authors recommend targeting a serum calcium level that is in the low-normal range and not normalizing the serum PTH concentration, to avoid the risk of hypercalcemia and/or hypercalciuria.

Introduction

General

A. Definition

The term pseudohypoparathyroidism (PHP) was first introduced in 1942 by Albright to describe patients who presented with hormonal parathyroidism (PTHb-resistant hypocalcemia and hyperphosphatemia with characteristic skeletal and developmental changes).

Pseudohypoparathyroidism (PHP) is a group of rare, related and highly heterogeneous disorders characterized by end-organ resistance to the action of parathyroid hormone (PTH). PHP and related disorders are caused by genetic and/or epigenetic changes resulting in downregulation of a cyclic adenosine mono phosphate (cAMP) generator, primarily linked to the GNAS gene [1].

The two main subtypes of PHP are types Ia and Ib (PHP-Ia, PHP- Ib) and are caused by molecular alterations within or upstream of the GNAS gene [1]. Multiple transcript variants encoding different isoforms have been found for this gene. Mutations in this gene result in pseudohypoparathyroidism type 1a, pseudohypoparathyroidism type 1b, Albright hereditary osteodystrophy, pseudohypoparathyroidism, McCune-Albright syndrome, progressive bone heteroplasia, polyostotic fibrous dysplasia of bone, and some pituitary tumors [2-5].

Methodology

Goals

a) Primary objective

To comprehensively update the description of pseudohypoparathyroidism epidemiologically, clinically, diagnostically and therapeutically [5-10].

b) Secondary objectives

- Identify data on the prevalence and incidence of PPHT globally and according to geographical areas
- Describe the basics and review new developments on the pathophysiological mechanisms of PPHT,
- Describe the clinical elements of PPHT,
- Evaluate PPHT diagnostic tools
- Describe the treatment of pseudohypoparathyroidism.

Materials and Methods

A criteria-based narrative systematic literature review PRISMA-P [11] (Annex 1) has summer carried out in leaning methodologically on there publication of the article of Zaugg and al [12] as well as the guide to recommendations relating to reviews systematic narrative of the Economic and Social Research Council (ESRC) [11-16].

There review of there literature has summer performed In THE basics of data international MEDLINE and Cochrane data base of systematic reviews by their search engine (respectively Pub Med). The data French speakers were searched with the search engines of the Database of the Scientific Literature in Health site (LiSSa), of the University Documentation System (SUDoc), the Catalog and Index sites Medical of language French (CISMeF). Gray literature was also integrated using Google Scholar. THE results have summer enriched by there reading of the references bibliographies of selected publications.

For each engine research, different equations have summer proposed in order to to arrive at the most sensitive result possible without neglecting the criterion of specificity of the results researches. The international review was carried out using the words of the Medical Subject Heading (MeSH) assigned to general medicine. These terms were then combined by THE bias of operators booleans (and, or). MEDLINE has SO summer Interrogates Thus by her engine of research Pub Med, the algorithm obtained was:

Epidemiological Data

Few data are available regarding the epidemiology of PPH in a well-defined population. Their prevalence is estimated at approximately 0.79 per 100,000 (according to Orphanet Report Series, November 2008). period prevalence of diseases was 3.4 (2.6-4.2) per million population in pseudohypoparathyroidism (95% confidence intervals [11]. Data for Africa are missing.

Physiopathological Mechanisms

Indirect evidence eliminates a defect in renal alpha-hydroxylase as a determining factor in this condition. Likewise, the increase in the size of the average cross-sectional area of periosteocyte lacunae, determined on decalcified bone sections obtained by Trans iliac biopsy, shows that osteocytes are sensitive to endogenous PTH, rejecting the response of cAMP to PTH in bone as a prerequisite for the action of PTH on bone.

The authors conclude from these data and previous experiences that the defect in parathyroid function in this condition is probably related to the existence of an abnormal PTH molecule and/or metabolism and/or interaction with the receptor sites. Endocrine function has also been studied. A pre-diabetic state was revealed, as well as latent primary hypothyroidism (TRH test). Prolactin release could not be stimulated by TRH, levodopa, metoclopramide, chlorpromazine, and insulin hypoglycemia. The latter produced normal release of ACTH (as established by plasma cortisol levels) and GH, and perhaps a slow glucagon and gastrin response. There was a deficit in urinary concentration when restricting fluid intake. This was only partially corrected by ADH administration [17].

Clinical Tables

Signs of hereditary Albright osteodystrophy may include [3]

Term used to indicate a constellation of physical characteristics originally described by Albright1, including a round face, stocky habitus with short stature, brachydactyly, and ectopic ossification. Short bones are not present at birth and result from premature

closure of the epiphyses, resulting in a period of reduced growth. Although all bones tend to be short, the shortening is most marked acrally (i.e., in the hands and feet). Later, developmental delay was added as an additional characteristic. Obesity, especially early-onset obesity, and macrocephaly in relation to height could also be part of it.

Progressive Bone Heteroplasia [18]

POH is defined by the presence of ectopic ossifications that are progressive and extend deep into the connective tissue. Ossifications can lead to severe ankylosis of affected joints and focal growth retardation.

Acrodysostosis [19]

Acrodysostosis is defined as the association of severe brachydactyly, facial dysostosis and nasal hypoplasia. Brachydactyly typically affects all of the phalanges, metacarpals, and metatarsals, except the thumbs and halluxes. On radiographs, the epiphyses are conical in shape, bone age is advanced, and abnormalities may be present at birth or shortly thereafter [20].

Cognitive characteristics

Cognitive disorders have summer reported in 40–70% of patients with PHP1A and in 0–10% of patients with PPHP or POH, and they are rarely observed in patients with PHP1B and have variable prevalence in patients with acrodysostosis [21]. Cognitive performance studies were undertaken only in PHP1A and they showed reduced scores compared to peers [22], with a mean IQ of 85.9 and a reduction of 21.5 IQ points below a sibling. unaffected [23]. Patients with PHP1A were found to have impaired executive function, delayed adaptive behavioral abilities, and increased rates of attention deficit hyperactivity disorder [23]. A retrospective review of developmental milestones showed greater delay in language versus gross motor skills, with and a tendency to improve in late childhood [24].

Neurological and neuropsychiatric manifestations

may be related to function in addition to the role of Gsα in brain development [25], and other organic CNS alterations, including Chiari 1 malformation [26] or prolonged periods of hypocalcemia and in some patients with cerebral calcifications [27,28].

Diagnosis of Pseudohypoparathyroidis

Diagnoses of PPH and AHO are based on the combination of clinical and biochemical features that may vary depending on the patient's age and family history. Serum calcium, phosphorus, and parathyroid hormone levels should be checked together, as well as vitamin D 25OH levels. The presence of hypocalcemia, hyperphosphatemia, normal 25 hydroxy vitamin D, and elevated parathyroid hormone suggests pseudohypoparathyroidism. A synthetic PTH challenge test can be performed (Ellsworth-Howard test) but is not necessary for diagnosis. An electrocardiogram is probably appropriate if significant hypocalcemia is present to assess for QT prolongation and risk of arrhythmia. Patients with PHP1 should have biochemical tests annually, including PTH, calcium, phosphate, TSH, and urine calcium [29]. Monitoring for appropriate height and growth is important, and testing for growth hormone deficiency should occur even if height is normal [30]. Screening and treatment of other endo -crinopathies, such as hypogonadism, should be individualized [31-34].

Upper and lower extremity x-rays are recommended during the initial evaluation to screen for brachydactyly or other bony malformations. (48;50.48). In PHP patients with AHO, genetic testing is essential. Genetic testing and counseling provide information about likely disease manifestations and future complications. Additionally, genetic testing can help determine patterns of family inheritance and provide insight into the possibility of affected offspring.

Treatment

Curative treatment

Once diagnosed, PTH resistance should be treated with activated forms of vitamin D, for example calcitriol or alfacalcidol, to increase serum calcium levels and thereby reduce PTH levels. The authors recommend targeting a serum calcium level that is in the low-normal range and not normalizing the serum PTH concentration, in order to avoid the risk of hypercalcemia and/or hypercalciuria. PTH levels should be maintained at the upper limit or slightly above the reference range (eg, 50 to 150 pg/ml), because the distal renal nephron remains sensitive to PTH and can reabsorb calcium, thereby reducing the risk of hypercalciuria.

Vitamin D analogues may be started in infants when PTH increases (eg, 100–150 pg/ml), before hypocalcemia develops. Calcium intake should follow age-appropriate guidelines through a regular diet or supplements. Severe hyperphosphatemia may be treated with oral phosphate binders other than CaCO3, if necessary. Since cholecalciferol therapy helps increase calcium absorption in hypocalcemic patients [35]. The authors suggest maintaining serum 25(OH) vitamin D levels within the normal range. Proper management of PTH resistance to reduce the calcium phosphate product to less than 55 may reduce the development or worsening of calcifications in the lens and brain, but will of course have no effect on heterotopic ossification. Treatment of PTH resistance and functional hypoparathyroidism requires regular monitoring of serum levels of calcium, phosphorus, PTH, monitoring of urinary renal calcium excretion (< 4 mg/kg per day in adults). child) and renal function. Most patients with PHP1A are not at risk of developing renal calcifications [36] unless they are overtreated, thereby increasing the risk of developing hypercalciuria [37].

Patients with hypothyroidism due to TSH resistance should receive oral thyroxine and have their thyroid function assessed regularly. Patients with PHP and short stature or reduced growth velocity should be evaluated for growth hormone (GH) deficiency. Although the authors lack long-term data to formally recommend GH treatment in patients with PPH and short stature, short-term results and small series have provided encouraging results for patients [38,39]. Body mass index, to prevent the development of obesity and metabolic complications. Weight control can be very

difficult, because obesity results in part from decreased resting energy expenditure, and patients may not respond to the standard approach of calorie restriction. Currently, there is no specific treatment for heterotopic ossifications. Small ossifications usually do not progress and do not require treatment. Ossifications that cause pain and/or irritation can be surgically removed unless a large area of skin is involved [40,41], however necessary to evaluate the effectiveness of these drugs.

Regular mobilization of the limbs and physiotherapy are necessary when ossification surrounds the joints. [42]. Future innovative therapies for patients with PPH may include phosphodiesterase inhibitors [43].

Differential diagnosis [44]

Pseudohypoparathyroidism manifests as hypocalcemia, hyperphosphatemia, elevated PTH levels, normal 25-hydroxyvitamin D, and normal renal function. Additionally, many patients express the Albright hereditary osteodystrophy phenotype, but not always. Although no other disease presents identically, many conditions share similar characteristics providing the following differential diagnoses.

True hypoparathyroidism

He has hypocalcemia and hyperphosphatemia, but PTH levels are low. True hypoparathyroidism is generally classified as surgical or non-surgical. The most common cause of hypoparathyroidism is post- parathyroidectomy surgery for the treatment of hyperparathyroidism or accidental removal of the parathyroid glands during other neck surgeries such as thyroidectomy. Nonsurgical hypoparathyroidism can result from a variety of inherited and acquired disorders. Examples include hemochromatosis or other infiltrative diseases and mutations in the PTH gene, the CaSR gene, and autoimmune polyendocrine syndrome type 1 (APS1), which presents with the triad of hypoparathyroidism, primary adrenal insufficiency, and cutaneous candidiasis. -mucosa.

Vitamin D deficiency

This condition presents with hypocalcemia and high levels of PTH (secondary hyperparathyroidism) but obviously low levels of vitamin D 25 OH.

Worsening of chronic kidney failure (CKD)

Patients with impaired glomerular filtration rate (GFR) experience similar results. The kidney is the primary site of 1-alpha-hydroxylation of 25OH vitamin D which produces the active form of the hormone 1,25(OH)2 vitamin D. Impairment of conversion to active vitamin D leads to its low levels in patients with CKD. In addition to low levels of active vitamin D, higher than normal phosphate levels frequently accumulate in the blood due to lower GFR. Low levels of active vitamin D and high levels of phosphate lead to increased parathyroid hormone release.

Prognosis

PHP's prognosis is variable. In mild forms of the disease, when treated appropriately with calcium and vitamin D, a normal life expectancy is not unreasonable. For others with more severe expression of the AHO phenotype, the presence obesity, sleep apnea, and mental retardation can cause significant morbidity and mortality [45].

Complications

Chronic hypocalcemia and hyperphosphatemia can lead to parkinsonism which may or may not resolve with appropriate calcium and vitamin D supplementation [46]. Children with brachydactyly may suffer from difficulties with fine motor skills. There is an increased prevalence of carpal tunnel syndrome in AHO patients [47]. There are reports of greater than normal spinal stenosis, as well as anterior presentation of spinal stenosis, which can lead to lower extremity paraparesis [48]. Finally, there is more than four times the risk of sleep apnea reported in childhood [49].

Deterrence and Education of Patients

Unfortunately, PHP is an inherited disorder, and once an individual has developed the condition, there is no cure. Genetic testing and counseling are recommendations for people with PHP, particularly if they are planning to have children, so that they can fully identify their risk of passing on the disease. Patients with PHP should have annual screening exams, even if they feel well.

Improving Health Care Team Outcomes

Patients with pseudohypoparathyroidism should be followed by an endocrinologist and neuropediatrician. Regular testing of serum calcium levels is necessary, as well as screening for other hormonal resistance. The management of each patient requires individualization to their specific expression of hormonal resistance.

The diagnosis, treatment, and management of pseudohypoparathyroidism requires an interprofessional health care team approach. The patient's family physician will seek the services of an endocrinologist, as noted above. A genetics specialist is also a necessary consultation. Participation of other members of the health care team will vary depending on the individual patient presentation. If intravenous interventions are necessary, the nurse will administer them and monitor the effectiveness of the treatment, reporting findings to the treating clinician. The pharmacist will prepare the IV and, in the case of oral treatment, can advise the patient on dosage and administration. As treatment progresses, the team should be able to adapt based on the patient's response and coordinate with the next steps outlined above in the Treatment section. Through open communication and a collaborative effort, the interprofessional team can help achieve the best possible outcomes for the patient. [Level 5].

Conclusion

Pseudohypoparathyroidism is characterized by resistance to parathyroid hormone (PTH) and therefore this condition mimics hypoparathyroidism with hypocalcemia and hyperphosphatemia. There are several subtypes depending on the signaling deficit associated with PTH action. Symptoms of this disease are similar

to those of hypoparathyroidism; clinical manifestations include: subcutaneous ossifications, brachydactyly, resistance to thyroid-stimulating hormone, short stature and early-onset obesity.

Molecular genetic and/or epigenetic testing is recommended. PTH resistance and secondary hyperparathyroidism should be managed to prevent hypocalcemia and increased bone resorption, respectively. The care is multidisciplinary for children and adults Treatment includes supplementation with calcium, 1,25-dihydroxycholecalciferol and vitamin D2.

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None.

Conflict of Interest

No Conflict of interest.

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