IDENTIFYING POTENTIAL CLASSIFICATION CRITERIA FOR CALCIUM PYROPHOSPHATE DEPOSITION DISEASE (CPPD): RESULTS FROM THE INITIAL PHASES


Background: Classification criteria for calcium pyrophosphate deposition disease (CPPD) will facilitate clinical research on this common crystal-related arthritis. ACR/EULAR are jointly sponsoring development of CPPD classification criteria using a multi-phase process.

Objectives: To report preliminary results from the first two phases of a four-phase process for developing CPPD classification criteria.

Methods: CPPD classification criteria development is overseen by a 12-member Steering Committee. Item generation (Phase I) included a scoping literature review of five literature databases and contributions from a 12-member Steering Committee. Item generation yielded 420 items (312 from the literature, 108 from experts/patients). The Advisory Groups, and an item rating exercise to assess the influence of Advisory Groups, and an item rating exercise to assess the influence of criteria development.

Results: Item generation yielded 420 items (312 from the literature, 108 from experts/patients). The Advisory Groups eliminated items they agreed were unlikely to distinguish between CPPD and other forms of arthritis, yielding 127 items for the item rating exercise. Fifty-six items, most of which had a modal rating of +/- 2 or 3, were retained for future phases (see Table 1). As numerous imaging items were rated +3, the Steering Committee recommended focusing on imaging of the knee, wrist, and one additional affected joint for calcification suggestive of CPP crystal deposition.

Conclusion: The ACR/EULAR CPPD classification criteria working group has adopted both data- and expert-driven approaches, leading to 56 candidate items broadly categorized as clinical, imaging, and laboratory features. Remaining steps for criteria development include domain establishment, item weighting through a multi-criteria decision analysis exercise, threshold score determination, and criteria validation.


DOI: 10.1136/annrheumdis-2021-eular.469

DUAL ENERGY CT HAS PROGNOSTIC VALUE IN GOUT BEYOND STANDARD CLINICAL MEASURES: A BEST EVIDENCE SYNTHESIS

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Background: Dual Energy CT Scan (DECT) can detect monosodium urate deposits in multiple tissues. EULAR gout guidelines (Richette, 2020) recognized the value of DECT in making a clinical diagnosis when joint aspiration is difficult. DECT shows crystal deposits in almost 50% of gout patients without tophi (Dalbath, 2017). Tophi are known to predict all-cause mortality (Vincent 2017, Perez-Ruiz 2013) and it is plausible that DECT could as well. A prognostic measure should be reliable and valid. DECT validity would be evident for death, disability and distress.

Objectives: This study used a best evidence synthesis approach to synthesize the evidence for DECT as a prognostic measure in gout.

Methods: PUBMED and EMBASE were searched from initiation to December 2019; keywords (Dual Energy Computed Tomography OR DECT, gout,
Disclosure of Interests: Sally Stauder: None declared, Paul M. Pelosi Shareholder of: Horizon Therapeutics plc, Employee of: Horizon Therapeutics plc.

DOI: 10.1136/annrheumdis-2021-eular.872

PREVALENCE OF HYPOPHOSPHATASIA IN ADOLESCENT RHENATOMY PATIENTS SCREENED BY LOW LEVELS OF ALKALINE PHOSPHATASE

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Background: Hypophosphatasia (HPP) is a genetic disease caused by one or more mutations in the alkaline phosphatase (ALP) gene, which encodes tissue-specific ALP and affects the mineralization process. Accordingly, arthralgia, fractures, and dental abnormalities have been reported in adults, and fatal cases in children. This metabolic disorder is commonly misdiagnosed with other more prevalent bone diseases due to its low prevalence and lack of recognition (i.e. chondrocalcinosis). However, no epidemiological studies on the prevalence of HPP in the rheumatological patient population have been available to date.

Objectives: To identify the prevalence of HPP in rheumatological patients screened for persistent low levels of ALP and association with mutations in the ALP gene.

Methods: All adult rheumatology patients were screened for pathological low levels of ALP (<35 IU/L) between January 1, 2017 and June 30, 2019 at the Department of Rheumatology, Clinic of Internal Medicine III, University Hospital Bonn, Germany. Medical files of patients with pathological low ALP levels were then reviewed for clinical signs and symptoms as well as results of genetic testing for HPP (full sequencing using Next Generation Sequencing).

Results: In total, 2,289 rheumatology patients were screened for low ALP levels. In 60 patients (2.62%), pathological low ALP levels were identified, while in 30 of these (1.31%), persistent low ALP levels were detected. In 19 of the 30 patients, genetic tests for ALP gene mutations were done. Seven out of 19 patients (36.84%) had HPP-related symptoms (fracture, dental abnormalities) with normal bone densitometry, while four of these patients (21.05%) had a history of fracture and three patients (15.78%) showed dental abnormalities. In addition to the typical HPP signs and symptoms, 13 patients (68.42%) showed mutations in the ALP gene. One of the ALP mutations was found to be a novel genetic variant, classified as pathological. Interestingly, no association with chondrocalcinosis was detected.

Conclusion: In summary, it can be concluded that HPP is an under-diagnosed condition with a higher proportion of affected rheumatological patients than previously thought (at least 0.56% of rheumatological patients vs. 0.01% in a Spanish healthy population). If we replicate these numbers for the German population (83 million, 5% of whom suffer from rheumatic conditions) the yield is approximately 4.15 million. This possibly indicates that 23,240 potential cases of HPP are currently not diagnosed. Therefore, implementation of a protocol in clinical practice to prevent underdiagnosis of HPP and to treat this disease appropriately is essential.

REFERENCES: