THU0553 LOW ALKALINE PHOSPHATASE LEVELS: COULD IT BE HYPOPHOSPHATASIA?

C. Tornero[1], P. Aguado[1], S. García[1], J. Tenorio[2], P. Lapunzina[2], A. Buño[3], J.M. Iturzaeta[3], C. Plasencia[1], I. Monjo[1], A. Balsa[1]. *[1]Rheumatology; [2]Genetics; [3]Clinical Laboratory, la Paz University Hospital, Madrid, Spain*

Background: Hypophosphatasia (HPP) is a rare inherited disorder of bone and mineral metabolism, caused by mutations in the ALPL (alkaline phosphatase liver type) gene, with reduction of activity of the tissue-non-specific isoenzyme of ALP (TNSALP). The clinical presentation is variable and adult forms of the disease are usually milder than those affecting infants and children, easily overlooked or misdiagnosed, which can lead to erroneous therapeutic decisions.
Objectives: The primary objectives of our study are to estimate the prevalence of patients with adult forms of HPP in a group of patients with persistent hypophosphatasemia, to analyze their clinical and functional characteristics and to compare these findings between those presenting or not these mutations.
Methods: In this Cross-sectional study, 1.536.711 ALP measurements owing to 386.356 patients were evaluated during a six-year period (2009–2015). Patients having at least two values below 35 IU/l and none above 45 IU/l constituted the study population. In total, 427 patients were included. Among them, 31 patients were excluded because of presenting causes of secondary HPP and 13 because of lost to follow-up. 108 patients were contacted by phone to fulfill a questionnaire about manifestations related to HPP and health assesment and in order to obtain blood samples to perform the genetic test.
Results: Demographic and clinical characteristics of both groups are shown in Table 1. Of the 108 patients evaluated, the genetic test results of 39 patients are available at this moment (the rest of the results are currently pending). 59% (23/39) tested positive for the genetic mutation.Despite data are still partial and although the results did not achieve statistical significance, we observed with a greater relevance a higher proportion of patients with HPP presenting with chronic bone pain (48,7% vs 25,6%,p=0,157), muscle weakness (15,4% vs 2,6%, p=0,112) and more necessity of analgesic medication (p=0,107) in patients with HPP in comparison with the control group. Furthermore, there was a non-significant trend in the HPP-group to present dental abnormalities and premature dental loss (7,6%), absent manifestations in the control group (p=0,123). In addition, orthopedic surgery was more common in the HPP group (12,8%) compared with the other group (0%), p=0,04. The prevalence of stress fractures was also higher in patients with HPP (7,69%). No significant differences were found in demographic characteristics, vertebral fractures, calcific periarthritis, condrocalcinosis, or FRAX index. In terms of biochemical tests, serum phosphate levels were higher in the HPP group (4,18 mg/dl) in relation to the control group (3,52 mg/dl), p=0,05. No differences were observed neither in pain assessment, measured with the Visual Analogue Scale nor in the Health Assessment Questionaire for disability between both groups.
Conclusions: The diagnosis of HPP can be difficult and is often missed or delayed, particularly in adults. The prevalence of HPP between patients with persistent low ALP is high and although the clinical presentation is milder in adults, it often presents with chronic bone pain, weakness, stress fractures and dental abnormalities. These data should promote a more proactive attitude towards detection of adult HPP.
Disclosure of Interest: None declared
DOI: 10.1136/annrheumdis-2017-eular.5547

Pts Characteristics	HPP group N= 23	Control group N= 16	P value
Demographic characteristics			
Mean age ± DS	52,78 ±13,28	49,13 ±10,70	p=0,366
Sex - Male - Female	 10/39 (25,6%) 13/39 (33,3 %)	 4/39 (10,2%) 12/39 (30,7%)	p= 0,236
Race - Caucasic - Black	 22/39 (56,4 %) 1/39 (2,56%)	 16/39 (41 %) 0/39 (0%)	p=0,398
Clinical characteristics			
Chronic bone pain, n/N (%)	19/39 (48,7 %)	10/39 (25,6%)	P= 0,157
Muscle weakness, n/N %)	6/39 (15,4%)	1/39 (2,6%)	P=0,112
Calcific periarthritis, n/N(%)	4/39 (10,25%)	1/39 (2,56%)	p=0,282
Condrocalcinosis n/N (%)	2/39 (5,12%)	0/39 (0%)	p=0,215
Patients presentig stress fractures, n/N (%)	3/39 (7,69%)	0/39 (0%)	
Vertebral fractures (n/N) (%)	2/39 (5,12%)	0/39 (0%)	p=0,193
Dental abnormalities and teeth loss n/N (%)	3/39 (7,69%)	0/39 (0%)	P= 0,123
History of orthopedic surgery	5/39 (12,8 %)	0/39 (0 %)	P=0,048
Use of analgesic medication for pain, n/N (%)	16 /39 (41,03%)	7 /39 (17,9%)	P=0,107
History of kidney disease	5/39 (12,8%)	2/39 (5,12%)	P=0,384
Biochemical data			
Alkaline phosphatase ± DS	21,6 ± 6,76	29,69 ± 3,44	P=0,0001
Serum calcium ± DS	9,34 ± 0,4	9,34 ± 0,4	P=0,994
Urine calcium ± DS	61, 47 ± 51,3		
Serum phosphate ± DS	4,18 ± 9,91	3,52 ± 0,48	P= 0,05
Urine phosphorus ± DS	341 ± 433		
Quality of life assessment			
VAS pain ± DS	3,86 ± 2,5	2,71 ± 3,1	P= 0,221
HAQ-DI ± DS	0,23 ± 0,36	0,2 ± 0,43	P= 0,764

THU0554 IMMUNOGLOBULIN G4-RELATED DISEASE IN HONG KONG – CLINICAL FEATURES, TREATMENT PRACTICES AND ASSOCIATIONS

P.H. Li, C. Ho, C.-S. Lau. *Department of Medicine, Queen Mary Hospital, Hong Kong, Hong Kong*

Background: Immunoglobulin (Ig) G4-related disease (IgG4-RD) is a systemic immune-mediated disease unifying what were previously considered to be unrelated individual organ disorders. The diagnosis and treatment of this characteristic fibroinflammatory condition continues to evolve, but generally remains an under-recognised disease. Local data outside Japanese and Caucasian populations is lacking and few studies have examined factors to predict disease severity or disease prognostication.
Objectives: We conducted this study to review the clinical features, treatment practices and factors associated with more extensive disease involvement in Hong Kong.
Methods: We retrospectively evaluated all patients with IgG4-RD over the past 13 years in our centre and combined this with patient data extracted from previous local publications. We analysed the clinical features, treatment practices and factors associated with the number of organ systems involved.
Results: One hundred and four patients (55 from our centre and 49 from literature review) were identified. Patients were predominantly older men (mean age 61.9 years, male:female ratio 3:1). Hepatobiliary and pancreatic (40.4%), salivary gland (33.6%), lymph nodes (29.8%) and eye (19.2%) were the most common systems involved. Lymphadenopathy was associated with glucocorticoid use (OR=2.65, p=0.034). Over 90% of patients had a serum greater than 135 mg/dl and a IgG4/total IgG ratio greater than 8%. Pre-treatment serum IgG4 levels correlated with the number of organ systems involved (β=0.347, p=0.004), and specifically with salivary gland involvement (mean 1109 mg/dl vs. 599 mg/dl, p=0.012).
Conclusions: We identified pre-treatment serum IgG4-RD to be associated with multi-system disease, especially with salivary gland involvement, highlighting the potential for its use in disease prognostication and monitoring. The reason for this particular correlation remains uncertain but highlights the importance of screening for salivary gland involvement in all IgG4-RD patients, especially in the presence of higher serum IgG4 levels. We also describe the clinical features and treatment modalities of the largest cohort of IgG4-RD in Hong Kong thus far. Increased physician awareness and multidisciplinary efforts are required for optimal management of this masquerading disease. Future studies, especially focusing on treatment strategies within the contexts of different epidemiology and patient characteristics, are urgently needed.
Disclosure of Interest: None declared
DOI: 10.1136/annrheumdis-2017-eular.1120

THU0555 THE RELIABILITY OF TWO VASCULITIS ACTIVITY SCORES (BIRMINGHAM VASCULITIS ACTIVITY SCORE AND BEHCET'S DISEASE CURRENT ACTIVITY FORM2006) IN ANTICIPATING DAMAGE IN A GROUP OF PATIENTS DIAGNOSED WITH BEHCET'S DISEASE

C.G. Buzatu, S.E. Daia-Iliescu, I. Saulescu, A. Borangiu, T. Gudu, V. Bojinca, D. Predeteanu, A. Balanescu, R. Ionescu, D. Opris-Belinski. *Internal Medicine and Rheumatology, "Saint Mary" Clinical Hospital, Bucharest, Romania*

Background: Validated disease activity scores and damage measurements were developed over time in order to allow a better way to evaluate patients and decide treatment plans.There are scores designed for a great variety of vasculitis like Birmingham Activity Score and others that are more specific like Behcet's Disease Current Activity Form2006.
Objectives: To evaluate the ability of the activity scores (BVASv3and BDCAF)to predict damage,and the influence of immunosuppressive therapy on damage progression,as measured byVDI, in a group of patients with Behcet's Disease.
Methods: A study was performed on a cohort of patients diagnosed with Behcet's Disease under surveillance in one tertiary Rheumatology Centre, from a non-endemic area.All documented cases of Behcet's Disease have been diagnosed according to The International Criteria for Behçet's Disease.The Birmingham Activity Score (BVAS)v3, Behcet's Disease Current Activity Form2006 (BDCAF)and Vasculitis Damage index (VDI) were calculated for all patients. Spearman's correlation coefficients were calculated between BVASv3 Score, BDCAF, VDI and immunosuppressive treatment.WindowsExcel/SPSS20.0 has been used to analyse the data.
Results: 20 patients were included in the study, with ages at the time of the