SAT0190 HFE GENOTYPE IN PATIENTS WITH RHEUMATIC DISEASES

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Background Hereditary haemochromatosis is a very common autosomal recessive disorder of iron metabolism. Among Northern Europeans the carrier frequency is estimated 1 in 10, while up to 1 in 200 are affected by the disease. Arthropathy is one early clinical manifestation of this disease (the spectrum ranged from arthralgia to classic polyarthrits), but the articular features are often misdiagnosed. We tested the usefulness of measuring the HLA-linked haemochromatosis gene (HFE) in rheumatology clinic population.

Objectives

Methods Over a period of 12 month 203 consecutive patients, mean age 62.0 years (33 male/170 female) attending our rheumatologic clinic were screened for HFE mutations (C282Y and H63D). Mutations were evaluated by separation on PAGE of digest (by *Sna*I and *Bcl*-I, for C282Y and H63D, respectively) of PCR-amplificates of DNA obtained from PBMC's.

Results The C282Y and H63D allele frequencies were 6.9 and 18.2 in patients with rheumatic diseases. Five patients were homozygote for H63D(2.5%), 1 for C282Y(0.5%). Three patients were compound heterozygotes (1.4%). Diagnosis of patients are summarised in Table 1. The observed C282Y allele frequency in rheumatic patients with undifferentiated arthritis exceeded that of healthy subjects or patients with chronic hepatitis C in Austria.¹

Abstract SAT0190 Table 1

	C282Y/WT	H63D/WT	H63D/H63D	C282Y/C282Y	WT/WT
Osteoarthritis	14	21	4	0	82
Undifferentiated	0	0	0	1	5
Rheumatoid	1	12	1	0	47
Psoriatic	1	4	0	0	6
Collagenosis	0	0	0	0	3
Temporal	0	0	0	0	1
Total	16	37	5	1	144

Distribution of HFE Gene Mutation in 203 rheumatologic patients.

Conclusion Determination of HFE genotype is clinically useful in the group of patients with undifferentiated peripheral arthropathy and may lead to identification of otherwise unsuspected C282Y homozygotes.

REFERENCE

- 1 Kazemi, et al. *Gastroenterology* 1999;**116**(1):12-34

SAT0191 QUANTITATIVE ULTRASOUND OF THE CALCANEUS IN PORTUGUESE CHILDREN AND ADOLESCENTS

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Background

Objectives The aim of this study was to determine value range of the parameters SOS, BUA and stiffness for Portuguese children and adolescents in age, sex and race groups.

Methods The study included 473 healthy children coming from two Lisbon schools. The population was 92,2% caucasian. There were 261 (55,2%) girls and 212 (44,8%) boys. Mean age was 10,7±2,4 year, range 6-17 yr. The Sahara Clinical Sonometer of Hologic was used to performed a quantitative ultrasound (QUS) measurement of the non-dominant calcaneus. Patient examination was performed by two rheumatologists.

Results The mean of BUA, SOS and stiffness were: 70.07±19.82 db/MHz, 1563.64±26.65 m/s and 98.96±17.7.

There were no statistical differences between sex or race in each groups.

We observed an increased BUA, SOS and stiffness from 6 to 9 yr in caucasian boys and then stabilised. At this time the means were respectively 81.06 db/MHz, 1582 m/s and 110.86. In caucasian girls the increase of values occurred two years later and until 14 yr. The Spearman's rho correlation coefficient shown, in caucasian girls, a influence of age, height and weight on QUS parameters. In boys, this factors affect only SOS and stiffness. There were no correlation with this factors in black.

Abstract SAT0191 Table 1

	Caucasian Girls (n = 240)	Caucasian Boys (n = 196)	Black Girls (n = 21)	Black Boys (n = 16)	Total n = 473
BUA	69.14 ± 18.49	70.12 ± 20.44	76.99 ± 26.07	74.21 ± 21.85	70.07 ± 19.82
SOS	1561.71 ± 24.16	1563.65 ± 27.83	1569.23 ± 39.42	1584.93 ± 17.77	1563.64 ± 26.65
Stiffness	97.65 ± 15.55	99.20 ± 19.26	103.95 ± 25.23	109.25 ± 14.99	98.97 ± 17.77
BMD	0.541	0.551 ± 0.122	0.581 ± 0.159	0.614	0.549 ± 0.112

Conclusion Measuring bone mass of children by QUS is good technique. However reference data ranges must be developed for each population.

SAT0192 RELATIONSHIP BETWEEN CLINICALLY DETECTED JOINT SWELLING AND EFFUSION DIAGNOSED BY ULTRASONOGRAPHY IN TALOCRURAL JOINTS IN PATIENTS WITH RHEUMATOID ARTHRITIS

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Background In clinical investigation it is often difficult to know if there is effusion in talocrural (TC) joint or not.

Objectives To compare the relationship between clinically detected swelling and effusion diagnosed by ultrasonography (US) in TC joints in patients with rheumatoid arthritis (RA).

Methods Thirty-one consecutive patients with RA entered the study. Twenty healthy persons formed a control group. In clinical assessment the TC joint was evaluated on the scale: normal or swelling. The US of the same joints was carried out. Altogether 62 TC joints of the RA patients and 40 of those of the controls were studied. All the clinical assessments were performed by one