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ABSTRACT BOOK

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Affiliated Scientific Societies

SIE (Italian Society of Hematology, *www.siematologia.it*)
AEHH (Spanish Association of Hematology and Hemotherapy, *www.aehh.org*)
SETH (Spanish Society of Thrombosis and Hemostasis, *www.seth.es*)
SIES (Italian Society of Experimental Hematology, *www.sies.ws*)
SISET (Italian Society for Studies on Hemostasis and Thrombosis, *www.siset.org*)
AIEOP (Italian Association of Pediatric Hematology/Oncology, *www.aieop.org*)
EAHP (European Association for Haematopathology, *www.socforheme.org/eahp*)
SIdEM (Italian Society of Hemapheresis and Cellular Manipulation, *www.emaferesi.it*)

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European Hematology Association (EHA)

EHA is a scientific society aiming to support research, education and clinical practice in hematology.
Its main objective is to be useful to scientific researchers, clinicians, medical students,
as well as all those working in other fields but who are interested in hematology.

The European Hematology Association was founded in June 1992.
Today, EHA – with over 2700 active members from 95 countries – is a consolidated organization
that pursues a large and growing number of projects and programs.

EHA aims to promote

- Exchange and dissemination of knowledge and scientific information in the field of hematology.
- Education and training in hematology.
- Medical practice in the area of hematology and the position of hematology as medical discipline.
- Scientific research in hematology.
- Exchange of information for all European doctors, scientists and other professionals interested in hematology.
- A unified European training program in hematology in collaboration with European National Societies of Hematology.

In order to achieve these goals, EHA

- Maintains regular contacts and organizes meetings with all European National Societies of Hematology.
- Holds an annual scientific and educational congress in a major European city; European Cooperative Groups and Networks are encouraged to take advantage of this major event to gather.
- Disseminates medical research, both basic and clinic, through the new journal Haematologica/The Hematology Journal.
- Has established a link with European National Societies of Hematology and other organizations such as the European Group for Bone Marrow Transplantation, European Association for Hematopathology, European Society of Medical Oncology, and American Society of Hematology.
- Provides postgraduate education through the annual congresses, seminars, courses, workshops and meetings organized in collaboration with the European School of Haematology.
- Has a Fellowship/Grants Program to promote research in hematology.
- Accredits scientific meetings and provides CME accounts in collaboration with the European National Societies for hematology.

If you recognize the need for a strong European Hematology Association
and would like to take advantage of the various activities of the Association,
you may wish to become a member of the EHA and contribute to its objectives.

Benefits of EHA Membership

- Subscription to Haematologica/ The Hematology Journal, including on-line access
- Reduced registration fee for EHA Annual Congresses
- Eligible to EHA Research Fellowships & Grants
- EHA Newsletter
- Access to EHA membership database
- Access to webcast sessions of the EHA Annual Congress – NEW!
- Entitled to apply for a scholarship to attend ESH-EHA scientific workshops

www.ehaweb.org

It was not identified in any of the 25 patients with CML and 30 normal ethnic Omani blood donor controls. Statistical analysis of the subgroups with ET and PRV patients between those who were JAK2 positive and negative did not reveal any discriminating variables, especially the platelet count and the hemoglobin levels respectively. However, In the PRV group who were on regular phlebotomy [n=14], 93% were JAK2 positive. *Summary and Conclusions.* A single acquired mutation of JAK2 was noted in almost half of the patients with myeloproliferative disorders.[48%] The incidence of JAK2 V617F mutation in myeloproliferative disorders from the Sultanate of Oman is similar to that reported by other groups. Early screening of suspected PRV patients for JAK2 V617F mutation rapidly identifies nearly all those patients who will ultimately need definitive treatment without invasive investigations.

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HOLOTRANSCOBALAMIN (HOLO-TC) FOR DIAGNOSING EARLY VITAMIN B12 DEFICIENCY

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Background. Vitamin B12 deficiency is a frequent problem, particularly among older persons. Early diagnosis of vitamin B12 deficiency is crucial because of the latent nature of this disorder and the possible risk of irreversible neurological damage and hematologic diseases. The standard screening test for vitamin B12 deficiency, measurement of total serum vitamin B12, has limitations of sensitivity and specificity. For low concentrations of total vitamin B12 there is likely to be misclassification of B12 status if relying on total serum B12 alone. Serum vitamin B12 bound to transcobalamin II (Holo-TC), constitutes only 6%-20% of total vitamin B12, and is the fraction of total vitamin B12 available for tissue uptake. Serum concentrations of Holo-TC has been proposed as a potential and alternative marker of early vitamin B12 deficiency. *Aims.*We investigated the usefulness of Holo-TC compared with total serum vitamin B12, in diagnosing of early vitamin B12 deficiency. *Methods.* We compared the performance of Holo-TC with total vitamin B12 to screen for metabolic vitamin B12 deficiency. The study included 54 serum samples from patients with concentrations of total vitamin B12 in the range of 150-300 pg/mL (gray-zone). Total serum vitamin B12 concentrations were determined by chemiluminescent microparticle immunoassay (CMIA) (Architet Abbott); serum Holo-TC concentrations were determined by microparticle enzyme immunoassay (MEIA) (Axsym Abbott). *Results.* Low levels of Holo-TC (<35 pmol/L) were observed in 19 samples (35%) with total vitamin B12 levels of 150-300 pg/mL. Linear regression analysis and Pearson correlation were used to analyse the association between the biochemical variables. Regression analysis shows that there is only a poor correlation between vitamin B12 and Holo-TC (r=0,495) for values of vitamin B12 falling in the gray-zone Figure 1.

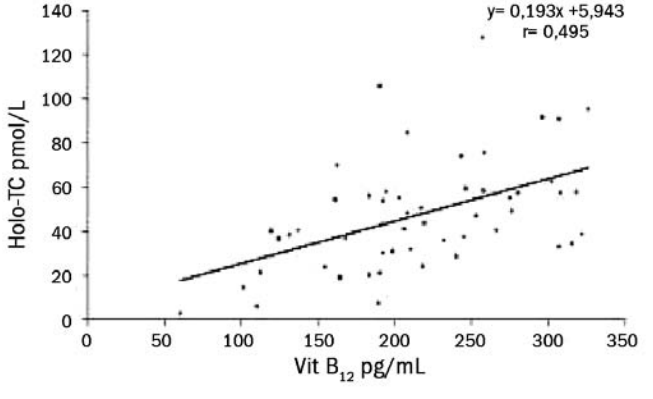

Figure 1.

Summary and Conclusions. Low values of Holo-TC was observed in a considerable number of patients with normal level of total vitamin B12.Since it can take months, even years, for a significant fall in total vitamin B12 levels, the more rapid decline in Holo-TC may be masked when measuring serum total vitamin B12. Therefore, total cobalamin concentration in the range of 150-300 pg/mL, cannot exclude a deficiency of the vitamin. Our results suggest that serum concentrations of Holo-TC is a more sensitive marker in diagnosing vitamin B12 deficiency when compared with serum total vitamin B12. In conclusion, our study demonstrated that Holo-TC can be used in screening studies as a first line parameter for detecting early deficiency before the development of clinical symptoms such as macrocytic anemia and neurological damages. Nevertheless, large scale clinical studies are warranted in order to clarify the usefulness of Holo-TC in the clinical setting.

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BETA-THALASSEMIA: PREVALENCE IN ALGARVE, PORTUGAL

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Background. Thalassemia is a genetic disease that makes part of an heterogeneous group of mendelian disturbs characterised by a diminution or absence in the production of one of the α or β Haemoglobin A chains (α2 β2). β-thalassemia is a variance of the thalassemic disease. This disease is found in many different ethnic groups and is more incident in some geographic areas, like Mediterranic zones where Algarve is insert. The migration can be an important factor on this disease prevalence once that occur immigration in Algarve provided from others geographic areas where β-thalassemia prevalence is high too. The detection of this disease is very important because is a way of prevention once that with this information it's possible to detect and inform the carriers, identify and give genetic counselling to the risk couples and an eventual prenatal diagnostic. *Aims.* The aim of this study is to determine the prevalence of this disease in population resident in Algarve (dependent variable), analysing its dependence of nationality and ethnicity (independent variables). *Methods.* The studied sample is formed by 28800 individuals, residents in Algarve. Data related to sex, age, nationality, ethnicity, and haemoglobinopathy type were collected between 1986 and 2006, among the National Program of the Haemoglobinopathies Control carried out by the Public Health Department of the Regional Health Administration of Algarve. Statistic data treatment was made with the program SPSS v. 15.0. *Results.* Most of the tested individuals are non carriers of any kind of haemoglobinopathy (90.7%) being β-thalassemia the most prevalent haemoglobinopathy in Algarve (7.5%) followed by Haemoglobin S (1.6%). This study allowed us to verify β-thalassemia as the most prevalent in people original from Algarve (9.3%), followed by the individual from other parts of the country (6.5%) and is higher in caucasians (8.1%) followed by black people (1.4%). *Summary and Conclusions.* Previous studies have shown that β-thalassemia presented values between 2.0% and 15.0% in Mediterranic areas. The result found in this study, 7.6%, shows the importance of this kind of study.

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SPECTRUM OF β-THALASSEMIA MUTATIONS IN THE KERMANSHAH PROVINCE OF IRAN

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Background. β-thalassemia is the most common single gene disorder in Iran. The presence of around 25000 affected individuals and two million carriers of β-thalassemia, requires an extensive study for the type of β-globin gene mutations in different ethnic groups of Iran. *Aims.* To find the spectrum of β-thalassemia mutations in the Kermanshah province of Iran with ethnic background of Kurd we studied 366 chromosomes from 183 unrelated homozygous β-thalassemia patients. *Methods.* Polymerase chain reaction (PCR), amplification refractory mutation system (ARMS) and direct sequencing were used to identify the type of β-thalassemia mutations. *Results.* As many as 20 different mutations (15 β0 and 5 β+ mutations) were identified. The most prevalent mutation was IVSII.1 G:A accounting for 33.3% mutations in patients. The frequency of the common mutations were [CD8/9 (+G), 13.7%], [IVSI.110 (G:A), 8.5%], [CD 36/37 (-T), 7.9%], [FSC 8 (-AA), 6%], [CD 15 (G:A), 4.9%], [IVSI.1 (G:A), 4.6%)], [IVSI.6 (T:C), 3.8%)], [CD 39 (C:T), 3%)], [IVSII.745 (C:G), 2.5%], [CD 44 (-C), 2.5%], [IVSI.5 (G:C), 2.2%], [IVSI, 3end(-25 del), 2.2%], [CD 83 (-G), 1.1%], [FSC 22/23/24 (-AAGTTGG), 1.1%]. Rare mutations were identified to be [IVSII. 2,3 (+ACGTTCTCTGAA), 0.6%], [IVSI.128 (T:C), 0.6%], [CD 6 (-A), 0.3%], [CD 37 (G:A), 0.3%], [CD 9/10 (+T), 0.3%]. The unknown alleles comprised 0.6% of the mutations. Around 82.5% of patients carried β0 type of mutations. *Conclusions.* The results of present study can help to establish prenatal diagnosis programs leading to lower medical cost.