### Blau syndrome--a chronic granulomatous, genetic disease

https://arctichealth.org/en/permalink/ahliterature80041

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<th>Author</th>
<th>Milman Nils</th>
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<td>Byg Keld-Erik</td>
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<td>Source</td>
<td>Ugeskr Laeger. 2006 Oct 16;168(42):3612-4</td>
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| Abstract | Blau syndrome is a rare hereditary granulomatous disease presenting in patients of young age with exanthema, granulomatous arthritis and uveitis. Genetic analysis has shown an autosomal dominant inheritance and a number of specific mutations on chromosome 16q in codon 334, of which the most predominant are R334W and R334Q. Blau syndrome exists in Caucasian, Asian and Afro-American families, and de novo mutations have been reported. The estimated minimum incidence in Denmark is 0.05 per 100,000 person-years. Blau syndrome has pathological, clinical and therapeutic features in common with sarcoidosis but rarely involves the lungs or other parenchymatous organs. Discrimination between Blau syndrome and early-onset sarcoidosis should rely on chromosome analysis. |

<table>
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<tr>
<th>PubMed ID</th>
<th>17069723 View in PubMed</th>
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Blood haemoglobin concentrations are higher in smokers and heavy alcohol consumers than in non-smokers and abstainers: should we adjust the reference range?

https://arctichealth.org/en/permalink/ahliterature91044

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Date: Jul-2009

Language: English

Publication Type: Article

Keywords: Adult
Age Distribution
Age Factors
Alcohol Drinking - blood - epidemiology
Body mass index
Cohort Studies
Denmark
Female
Ferritins - blood
Hemoglobins - analysis - standards
Humans
Male
Middle Aged
Questionnaires
Reference Values
Retrospective Studies
Sex Factors
Smoking - blood - epidemiology

Abstract: The blood haemoglobin concentration is one of the most frequently used laboratory parameters in clinical practice. There is evidence that haemoglobin levels are influenced by tobacco smoking. The objective of this study was to evaluate the impact of smoking and alcohol consumption on haemoglobin concentrations in apparently healthy subjects living at sea level. A retrospective, epidemiological cohort study was performed in 1984. Participants were 1,503 men and 1,437 non-pregnant women evenly distributed in age cohorts of 30, 40, 50, and 60 years. Information of smoking habits and alcohol consumption were obtained by a questionnaire. Haemoglobin was measured in the fasting state on Coulter-S. Men displayed no difference in mean haemoglobin levels in the four age groups. In women, mean haemoglobin increased gradually with age (p = 0.001). Fifty-nine percent of men and 50% of women were daily smokers. Female smokers displayed a significant positive correlation between number of cigarettes/day and haemoglobin concentrations (r = 0.12, p = 0.002). Heavy smokers (more than ten cigarettes/day) had significantly higher mean haemoglobin (1.4% higher in men, on average 3.5% higher in women) than non-smokers (p 14 drinks/week and more than seven drinks/week for men and women, respectively, increased mean haemoglobin by 1.3% in men and by average 1.9% in women compared with those consuming

PubMed ID: 19039534 View in PubMed
Cobalamin status during normal pregnancy and postpartum: a longitudinal study comprising 406 Danish women.

https://arctichealth.org/en/permalink/ahliterature82605

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Keywords: Adult
Cohort Studies
Denmark - epidemiology
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Hemoglobins - analysis
Homocysteine - blood
Humans
Infant, Newborn
Methylmalonic Acid - blood
Postpartum Period - blood
Pregnancy
Pregnancy Complications - epidemiology
Pregnancy outcome
Pregnancy Trimester, Third
Reference Values
Vitamin B 12 - blood
Vitamin B 12 Deficiency - epidemiology

Abstract: OBJECTIVES: To assess cobalamin (vitamin B(12)) status during normal pregnancy and postpartum in a longitudinal setting. METHODS: This study was performed in 1995-1996. It comprised 406 healthy, pregnant Danish Caucasian women, living in Copenhagen County. Cobalamin status, i.e. plasma (P-) cobalamin, P-methylmalonic acid and P-homocysteine was measured at 18, 32 and 39 wk gestation and 8 wk postpartum during lactation. RESULTS: P-cobalamin showed a gradual, significant decline during pregnancy (P

PubMed ID: 16548919 View in PubMed ✔


https://arctichealth.org/en/permalink/ahliterature83091
OBJECTIVE: To review the 13-year clinical experience of a single center's adult lung transplantation program.

METHODS: From January 1992 to December 2003, 369 lung transplantations were performed on 362 patients. Single lung transplantation was performed in 234 cases, double lung transplantation in 113 cases (comprising en-bloc double lung transplantation in 44 cases and bilateral sequential lung transplantation in 69 cases), heart-lung transplantation in 21 cases, and lobe of lung transplantation in 1 case. Recipient diagnoses included chronic obstructive pulmonary disease (COPD) (n = 175), alpha1 antitrypsin (alpha1AT) deficiency (n = 86), cystic fibrosis (n = 36), pulmonary fibrosis (n = 20), Eisenmenger syndrome and secondary pulmonary hypertension (n = 24), primary pulmonary hypertension (n = 8), sarcoidosis (n = 7), silicosis (n = 4), bronchiectasis (n = 1), and graft-vs-host disease (n = 1). RESULTS: For patients surviving to discharge, the median duration of the intensive care unit stay was 3 days (1-67), and the median duration of the post-operative hospital stay was 37 days (16-144). Mortality for the entire series was 6% at 30 days and 10% at 90 days. The main causes of post-operative inpatient death were primary graft failure (41%), sepsis (29%), cardiac (15%), and hemorrhage (9%). The 1-, 3-, 5-, and 10-year actuarial survival rates for the entire series was 81%, 68%, 63%, and 36%, respectively. There were no significant differences in survival between types of transplant. No significant differences in survival were seen between alpha(1)AT deficiency and COPD patients after stratifying for age. Cox regression analysis demonstrated that age 60 years or older, donor age 50 years or older, and a recipient pre-operative body mass index of 25 or higher were independent predictors of poor survival. CONCLUSIONS: This center has 1-, 3-, and 5-year survival rates comparable to other high volume centers. Recipient age, pre-operative body mass index, and donor age significantly influence outcome after lung transplantation.
Frequencies of the haemochromatosis gene (HFE) variants C282Y, H63D and S65C in 6,020 ethnic Danish men.

https://arctichealth.org/en/permalink/ahliterature93033

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Date: Sep-2008

Language: English

Publication Type: Article

Keywords: Amino Acid Substitution
Blood Donors
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Ethnic Groups - genetics
Europe
Gene Frequency
Genetic Variation
Histocompatibility Antigens Class I - genetics
Humans
Infant, Newborn
Male
Membrane Proteins - genetics
Polymorphism, Single Nucleotide

Abstract: The objective was to assess the frequencies of haemochromatosis (HFE) gene mutations or variants C282Y, H63D and S65C in ethnic Danes. This is a prospective epidemiologic population study. A cohort of 6,020 Danish men aged 30-50 years was screened for HFE C282Y (c845G-->A), H63D (c187C-->G) and S65C (c193A-->T) gene variants, assessed on saliva or blood samples by restriction fragment length polymorphism (RFLP) analysis. The C282Y gene variant allele was present in 5.6%, H63D in 12.8% and S65C in 1.8% of the chromosomes. In the entire series, we observed 1.4% H63D/C282Y, 0.1% S65C/C282Y and 0.4% H63D/S65C compound heterozygotes. The C282Y allele frequency in Denmark is of similar order as reported in other Scandinavian countries: Iceland 5.1%, Faeroe Islands 6.6%, Norway 6.8% and Sweden 5.8%. Also, the H63D frequency in Denmark is close to the frequencies in other Scandinavian countries: Iceland 10.9%, Faeroe Islands 15.2%, Norway 11.4% and Sweden 12.1%.

PubMed ID: 18542962 View in PubMed
Health-related quality of life in adult survivors of childhood sarcoidosis.
https://arctichealth.org/en/permalink/ahliterature90358

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Source: Respir Med. 2009 Jun;103(6):913-8
Date: Jun-2009
Language: English
Publication Type: Article
Abstract: AIM: To describe health-related quality of life (hrQOL) in adult subjects who had sarcoidosis in childhood.
METHODS: Forty-six children (24 boys), all ethnic Danes

PubMed ID: 19155167 View in PubMed

Hemoglobin concentrations in 358 apparently healthy 80-year-old Danish men and women. Should the reference interval be adjusted for age?
https://arctichealth.org/en/permalink/ahliterature93592

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Date: Feb-2008
Language: English
Publication Type: Article
Abstract: BACKGROUND AND AIMS: In elderly Danes, reference intervals for hemoglobin (Hb) concentrations are derived from younger population groups. The aim was to examine reference intervals for Hb and cut-off limits for anemia by application of criteria for normality to a representative population of 80-year-olds. METHODS: Participants in this epidemiological health survey cohort were 358 subjects (171 men) 80 years of age. A dietary survey was performed in 232 subjects. Blood samples included Hb, red cell indices, serum ferritin, serum C-reactive protein, renal and hepatic function tests. Normality criteria for Hb were: 1) values in all participants; 2) values in apparently healthy subjects; 3) values in 10-year survivors. Hb was compared with muscle strength, physical performance and diet. RESULTS: In the entire series, median Hb was 140 g/L, 5-95 percentile 116-160 g/L in men, and 131 g/L, 5-95 percentile 114-147 g/L in women (por=20 had a lower frequency of anemia. There was no correlation between Hb and dietary or supplemental iron intake. In men, Hb was correlated to meat consumption. CONCLUSIONS: WHO decision limits for anemia should not be lowered in 80-year-old subjects. "Optimal" Hb concentrations with respect to survival appear to be at least 140 g/L in men and 131 g/L in women. Further research should evaluate whether not only treating anemia, but also increasing Hb by using erythropoietin and hematinsics, may improve functional status and survival in the elderly.

PubMed ID: 18283222 View in PubMed
Higher prevalence of anemia among pregnant immigrant women compared to pregnant ethnic Danish women.

https://arctichealth.org/en/permalink/ahliterature77782

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Source: Ann Hematol. 2007 Sep;86(9):647-51

Date: Sep-2007

Language: English

Publication Type: Article

Abstract: The aim of the study was to investigate whether the well-known high anemia prevalence in pregnant women from the eastern Mediterranean and Asian regions decreased when the women immigrated to a low-frequency region (Denmark). During 70 months, 1,741 pregnant immigrant women referred from primary care to an obligatory hemoglobinopathy screening were eligible for the study, as their screening was negative. To compare this group with a cohort of 205 pregnant ethnic Danish women, the groups were matched by gestational age, and a total of 406 immigrant women were included. Hemoglobin (Hb) and iron status parameters were examined in the two groups. The prevalence of anemia was higher in the immigrant group (20.0%) compared to the Danish women (4.9%) (P

PubMed ID: 17486340 View in PubMed

Reference intervals for haematological variables during normal pregnancy and postpartum in 434 healthy Danish women.

https://arctichealth.org/en/permalink/ahliterature77315

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Date: Jul-2007

Language: English

Publication Type: Article
AIM: To report reference intervals for haematological variables during normal pregnancy and postpartum.

MATERIAL AND METHODS: The series comprised 434 healthy ethnic Danish women with a normal pregnancy > or =37 wk duration and a normal delivery with newborns weight >2500 g. Blood samples were obtained at 18, 32 and 39 wk gestation and at 8 wk postpartum. The following variables were analysed: Haemoglobin (Hb), haematocrit (Hct), blood erythrocyte count, mean corpuscular volume, mean corpuscular haemoglobin, mean corpuscular haemoglobin concentration, white cell count, platelet count, erythrocyte folate, plasma folate, plasma cobalamin, plasma methylmalonic acid, plasma total homocysteine, serum ferritin, serum soluble transferrin receptor and plasma creatinine. Reference intervals were calculated using log(10)-transformed values (which showed normal distributions) as mean +/- 1.96 x SD. RESULTS: The lower reference value for Hb during pregnancy was 6.45 mmol/L (105 g/L) and 7.3 mmol/L (118 g/L) postpartum. The lower reference value for Hct was 0.31 in pregnancy and 0.35 postpartum. There was a gradual decline in the lower reference value for erythrocyte folate during pregnancy and postpartum from 0.46 to 0.29 micromol/L and in plasma folate from 6 to 4 nmol/L. Lower reference value for plasma cobalamin declined during pregnancy from 96 to 71 pmol/L, but increased postpartum to 148 pmol/L. Upper reference value for plasma homocysteine increased gradually during pregnancy and postpartum from 11.0 to 20.6 micromol/L. Geometric mean serum ferritin at 18 wk gestation was 32 microg/L. Plasma creatinine values were low during pregnancy and displayed a significant increase postpartum. CONCLUSION: The characteristic changes occurring in haematological indices during pregnancy and postpartum are described in this study. The results may be used as reference values in the assessment of health status of pregnant women with a similar socio-economic and racial background.
Abstract: BACKGROUND: The biological function of rubidium (Rb) is unknown, but this alkali metal probably has a normal biologic role. OBJECTIVE: To measure the content of Rb in liver tissue samples from Greenlandic Inuit using X-ray fluorescence spectrometry, and compare the results with those obtained in liver samples from ethnic Danes. STUDY DESIGN: Observational, descriptive survey on environmental pathology. METHODS: The setting was related to forensic medicine and hospitalised care in Nuuk, Ilulissat and Copenhagen. Normal liver tissue was obtained at autopsy from 50 Greenlandic Inuit (27 men) with a median age of 61 years (range 23-83) and from 42 Danes (31 men) with a median age of 38 years (range 16-83). RESULTS: Liver Rb content in Inuit was not significantly different compared with Danes. There was no significant gender difference in liver Rb content either in Inuit or in Danes. The content of Rb given as median (5-95 percentile) was 0.1837mmol/kg dry liver (0.1041-0.3147) in Inuit, and 0.1965mmol/kg dry liver (0.0799-0.2815) in Danes (p=0.6). There was an inverse correlation between liver Rb content and age in Inuit (r(s)=-0.45, p=0.002) but not in Danes. Median hepatic Rb index (Rb content in micromol/kg dry weight divided by age in years) in Inuit was 3.05 and in Danes 4.21 (p=0.02). The correlations between liver Rb and liver potassium content were: Inuit r(s)=0.28, p=0.07; Danes r(s)=0.25, p=0.08; combined series r(s)=0.34, p=0.01. CONCLUSIONS: Inuit have liver Rb levels, which are quite similar to the levels found in Danes. In Inuit, liver Rb content appears to decrease with age.