



# FINAL PROGRAM AND ABSTRACTS

THE SEVENTH ISABS CONFERENCE IN FORENSIC, ANTHROPOLOGIC  
AND MEDICAL GENETICS AND MAYO CLINIC LECTURES  
IN TRANSLATIONAL MEDICINE



JUNE 20-24, 2011  
Bol, island of Brač  
CROATIA

[www.isabs.hr](http://www.isabs.hr) [info@isabs.hr](mailto:info@isabs.hr)

**STUDYING FREQUENCIES AND INTERACTION OF ALLELES C677T AND A66G IN DISTRIBUTION FOR COMPOUND GENOTYPES MTHFR/MTRR OF FOLATE CYCLE GENES IN EASTERN UKRAINE**Grechanina O<sup>1</sup>, Gusar V<sup>2</sup>

<sup>1</sup>Institute of Clinical Genetics, Kharkov, Ukraine; <sup>2</sup>Specialized Medical Genetic Centre, Kharkov, Ukraine  
v\_gusar@mail.ru

According to a pilot study (Matalon et. al., 2007), Ukrainian population may have a higher frequency (0.57) of allele 66G MTRR. Polymorphisms interaction C677T and A66G may lead to enhancement or compensation of phenotypic manifestations each of them individually. Thereby, we observed more extensive group of patients (1238) with inherited diseases and the polymorphisms, suggesting interaction on phenotype level. SNPs in MTHFR and MTRR were screened by allele-specific PCR. The gene fragments were visualized in 3% agarose gel. The being studied and expected frequencies of alleles and allele compounds of C677T MTHFR and A66G MTRR were calculated. Allele frequency 677T is 30.0% and allele frequency 66G is 57.0%. Difference between being studied and expected frequencies were observed for 2 from 9 compounds with confidence interval > 95 %. The being studied of compound frequency C677T Htzg/A66G Hmzg (17.0%) was higher than expected frequency (12.9±3.6%), and heterozygotes frequency C677T N/A66G Htzg (20.1%) was less than expected (25.8±4.0%). Compound C677T Htzg/A66G Hmzg may be maintained by natural selection due to mutual biochemical compensation of mutant alleles, since it is known, that decreased activity of MTHFR leads to low/normal level of folic acid, while MTRR is to high/normal level.

**Keywords:** folate cycle, MTHFR, MTRR, Ukraine, allele frequency

Presentation number: MG 5

Abstract number: ABS-130-ISABS-2011

**GENETIC AND EPIDEMIOLOGICAL CHARACTERISTICS ATOPICAL DISEASES AT THE KHARKOV POPULATION****Bezrodnaya AI<sup>1</sup>, Khodosh EM<sup>2</sup>, Kapelyshnaya YF<sup>3</sup>**

<sup>1</sup>V.N. Karazin Kharkov national university, Kharkov, Ukraine; <sup>2</sup>City pulmonology center of Kharkov, Kharkov, Ukraine; <sup>3</sup>Regional skin and venerology clinic of Kirovograd, Kirovograd, Ukraine  
*bezrodnaya@mail.ru*

To investigate correlation intersexual genetic and epidemiological parameters optimising clinical diagnostics of bronchial asthma (BA) and atopic dermatitis (AD). We have investigated 119 probands with BA from 46 (38, 7 %) men and 73 (61, 3 %) women at the age from 18 till 85 years; 297 patients with AD, from which 130 (43, 7 %) men and 167 (56, 3 %) women at the age from 0 till 80 years and older. The probability to be ill by the end of life was calculated as the cumulative diseasing (populations frequency). Was estimated angular transformation  $\phi$ ,  $\chi^2$  on a significance value 0, 05. From 119 observed probands with BA at 66 (56 %) patients was revealed skin signs atopy. Calculated population frequencies have shown that for men of the Kharkov population they make 3, 87 %, and for women - 3, 12 %, ( $p < 0, 05$ ); for BA at men - 7, 7 %, and at women - 8, 7 %, ( $p < 0, 05$ ). Investigating of frequency AD among patients with BA has shown that among men it has made 4, 6 %, and among women - 5, 2 % that is authentic above populations frequencies AD ( $p < 0, 05$ ). Presence the AD is regarded as the factor of high risk of development BA. Taking into account genetic heterogeneity of atopy the alleles dose predetermines development of its forms and disease severity level. Women in most degrees, than man's requires of formation of heavy currents.

**Keywords:** bronchial asthma (BA) , atopic dermatitis (AD), population, genetics, epidemiology