Topic: Preconception Screening

HIGH CARRIER FREQUENCY OF CFTR, PAH, GJB2 AND GALT GENES MUTATIONS ASSOCIATED WITH MONOGENIC DISEASES IN RUSSIAN POPULATION

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Objectives: *CFTR*, *PAH*, *GJB2* and *GALT* genes are responsible for autosomal recessive inherited diseases Cystic Fibrosis, Phenylketonuria, hereditary deafness and galactosemia respectively. Several frequent mutations are responsible for majority of disease cases but spectrum and frequencies of different genotypes may vary between different populations.

Methods: The aim of the study was to determine the frequency of common *CFTR*, *PAH*, *GJB2* and *GALT* mutations in healthy Russian population and optimal mutations spectrum for preconception screening. 2151 healthy persons were genotyped for 11 *CFTR*, 12 *PAH*, 1 *GJB2* and 1 *GALT* mutations. Kissing probes method was used for SNP detection.

Results: 45 carriers (2.1%) of *CFTR* mutations, 60 carriers (2.8%) of *PAH* mutations, 73 carriers (3.4%) of *GJB2* mutations and 12 carriers (0.6%) of *GALT* mutations were discovered. Only 4 persons had combination of 2 mutations so altogether we detected 186 carriers (8,6%) of mutations in *CFTR*, *PAH*, *GJB2*, *GALT* genes.

Conclusions: Study results can be used to choose optimal mutations spectrum for screening. Having regard to the frequencies of mutation carriers in Russian population preconception screening seems to be reasonable.

CFTR mutation	Heterozygote count
F508del	26
R117H	6
3849+10kbC>T	6
N1303K	3
dele2,3(21kb)	2
E92K	1
L138ins	1
W1282X, G542X, 1677delTA, 3944delGT	0
PAH mutation	Heterozygote count
R408W	49
IVS4+5G>T	3
R158Q	2
R261Q	2
IVS10nt546	2
Y414C	1
IVS12+1G>A	1
R252W, R261X, E280K, P281L, c.836C>T	0
GJB2 mutation	Heterozygote count
35delG	74
GALT mutation	Heterozygote count
Q188R	12