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PTH-101

ASSOCIATION OF ENDOTHELIAL NITRIC OXIDE SYNTHASE POLYMORPHISMS IN PRIMARY BILIARY CIRRHOSIS (PBC) PATIENTS AND FIRST DEGREE RELATIVES (FDR) OF CRETAN ORIGIN

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Introduction and background Twin and family studies suggest that there is a significant heritable component to PBC. It has been suggested that increased nitric oxide (NO) synthesis may contribute to the development of the hyperdynamic circulation observed in cirrhotic patients, while total NO circulating levels are enhanced in PBC.

Aims The authors examined two endothelial nitric oxide synthase (eNOS) gene polymorphisms, a 27-bp repeat in intron 4 and a Glu298Asp in exon 7, to assess a possible association with the development of PBC in a genetically homogenous Cretan population.

Methods Genomic DNA was extracted from 91 PBC patients (77 female), 82 AMA+, median age 68 years (41–84 years),

Table 1 PTH-101

eNOs intron 4 genotype	PBC (N=91)	FDR (N=101)	Controls (N=100)
Genotype bb	57 (62.6%)	69 (68.3%)	75 (75%)
Genotype ab/aa	34 (37.4%)*	32 (31.7%)**	25 (25%)
Allele a	40 (22%)	33 (31.2%)	29 (14.5%)
Allele b	142 (78%)	169 (68.8%)	171 (85.5%)
eNOs exon7 genotype	PBC (N=91)	FDR (N=101)	Controls (N=100)
Genotype TT	12 (13.2%)	12 (11.9%)	14 (14%)
Genotype GT	65 (71.4%)	61 (60.4%)	68 (68%)
Genotype GG	14 (15.4%)#	28 (27.7%)##	18 (18%)

*p=0.065, **p=0.293.
#p=0.682, ##p=0.259.

48/91 Ludwig stage I–II and 101 FDR (63 female), median age 41 (age range 18–80 years), 3/101 AMA+ and 20/101 ANA+ of Cretan origin. 100 sex, age and geography matched healthy controls (76 female), median age 69 years (39–86 years), 9/100 ANA+. Exon7 polymorphism was determined by PCR–restriction fragment–length polymorphism analysis. Intron4 polymorphism was determined using PCR amplification with oligonucleotide primers. Distribution of genotypes and alleles was compared by x^2 test or Fisher's exact test.

Conclusions In Cretan PBC patients both eNOs intron 4 VNTR and Glu298Asp in exon 7 eNOs polymorphism are not associated with disease occurrence or disease stage.

Competing interests None.

Keywords eNOS, primary biliary cirrhosis.

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