

Genes-4U

Interleukin – 6 promoter polymorphisms at -597, -572 and -174

Interleukin - 6 (IL6) plays key roles in **hematopoiesis, immune, and acute phase responses**.

Dysregulated IL6 expression is implicated in diseases such as **atherosclerosis** and **arthritis**. The interleukin-6 gene possesses promoter polymorphisms G-597A, G-572C and G-174C in partial linkage disequilibrium which affect the transcription of the interleukin-6 gene and the production of interleukin-6 protein. In vitro activity studies by transient transfection of promoter-reporter gene constructs into an endothelial-like cell line yielded the following haplotype / phenotype correlations (1) :

Haplotype (-597, -572 and -174)	Phenotype	Frequency (%)
GGG	High IL-6 production	54
GCG	Intermediate IL-6 production	5
AGG	Intermediate IL-6 production	0.3
AGC	Intermediate IL-6 production	40
GGC	Intermediate IL-6 production	0.6

Thus, in most cases **the G-174 allele is associated with high IL-6 production**.

A variety of clinical situations have been studied in relationship with the G-174 allele and the other polymorphisms in the IL-6 gene promoter (see reference table below).

References

(1) Terry CF, Loukaci V, Green FR. Cooperative influence of genetic polymorphisms on interleukin 6 transcriptional regulation. J Biol Chem 2000 Jun 16;275(24):18138-44 (PMID: 10747905)

Conditions associated with G-174C	PMID *
IL-6 after Coronary artery bypass graft (also with G-572C)	11557672
C-reactive protein (CRP)	11922913
CRP and bone resorption markers	12519862
Platelet and WBC count	11397324
Triglycerides and other lipid abnormalities	10720087
Energy expenditure and insulin sensitivity	12540635
Cortisol and hyperandrogenism (also with G-597A)	11889177
Recurrent aphthous stomatitis	12140751
Kidney allograft survival	12371985
Sepsis survival	11902285
Alzheimer's disease	12352619
Kaposi sarcoma	11001912
Antibodies to human hsp60	11862386
In vivo endothelial function	11895465
Multi-infarct dementia	12086705
Peripheral artery occlusive disease	12217290
Modulation of the effect of alcohol on carotid atherosclerosis	12574550

*** To view reference enter the PMID into the PubMed search window and press ENTER**