

Z610 Human RPS4X/Y genes and pseudogene family: chromosomal localization and phylogenetic analysis

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The human ribosomal protein S4 genes, RPS4X and RPS4Y, are located on the X and Y chromosomes. They have been postulated as candidate for Turner syndrome which is characterized by gonadal dysgenesis, short stature, and various external and internal anomalies. The RPS4X and RPS4Y proteins differ at 19 of 263 amino acids. Both genes are widely transcribed in human tissues. Using the BLAST search program, we identified sixteen RPS4 pseudogenes from the human genome and analysed them phylogenetically. The RPS4-C12-1, C12-2, and C12-3 pseudogenes from chromosome 12 have been evolved independently during hominid evolution. The RPS4X gene from X chromosome is closely related to the RPS4-C12-2 from chromosome 12 and RPS4-C5 from chromosome 5, whereas the RPS4Y gene is very closely related to RPS4-C16 from chromosome 16. The exact mapping of the RPS4 pseudogene family was performed, indicating that the RPS4 pseudogene family was mapped on human chromosomes 1, 2, 5, 6, 8, 10, 11, 12, 13, 16, 18, 19, and 20. Taken together, the precise chromosomal localization and phylogenetic relationship of the RPS4 pseudogenes could be of great use in further study for understanding the Turner syndrome.

Z611 Positive association of COMTmet/met with breast cancer

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Catechol-O-methyltransferase (COMT, EC 2.1.1.6) catalyzes the O-methylation for inactivation of catecholestrogens, which leads to oxidative DNA damage. The val→met at codon 158 of COMT encodes a thermolabile form of the enzyme with reduced activity, which results in accumulation catecholestrogens and participate in estrogen-induced carcinogenesis. We investigated that the association between the val158met mutation and the breast cancer risk. The COMT genotypes determined in 110 breast cancer patients and in 329 controls by PCR-NlaIII RFLP. The frequencies of COMTval/met and COMTmet/met genotype in breast cancer (46.4% and 10.0%) were higher than in controls (43.8% and 5.1%), respectively. And the odds ratio for COMTmet/met genotype was 2.3 [95% confidence interval (CI), 0.99-5.16] (p=0.05). These results suggest that COMTmet/met genotype positively associated with breast cancer risk.