

## ECHO.....

**Association between a functional polymorphism in the serotonin transporter gene and diarrhoea predominant irritable bowel syndrome in women**

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**Background and aims:** Serotonin (5-hydroxytryptamine, 5-HT) is an important factor in gut function, playing key roles in intestinal peristalsis and secretion, and in sensory signalling in the brain-gut axis. Removal from its sites of action is mediated by a specific protein called the serotonin reuptake transporter (SERT or 5-HTT). Polymorphisms in the promoter region of the SERT gene have effects on transcriptional activity, resulting in altered 5-HT reuptake efficiency. It has been speculated that such functional polymorphisms may underlie disturbance in gut function in individuals suffering with disorders such as irritable bowel syndrome (IBS). The aim of this study was to assess the potential association between SERT polymorphisms and the diarrhoea predominant IBS (dIBS) phenotype.

**Subjects:** A total of 194 North American Caucasian female dIBS patients and 448 female Caucasian controls were subjected to genotyping.

**Methods:** Leucocyte DNA of all subjects was analysed by polymerase chain reaction based technologies for nine SERT polymorphisms, including the insertion/deletion polymorphism in the promoter (SERT-P) and the variable tandem repeat in intron 2. Statistical analysis was performed to assess association of any SERT polymorphism allele with the dIBS phenotype.

**Results:** A strong genotypic association was observed between the SERT-P deletion/deletion genotype and the dIBS phenotype ( $p = 3.07 \times 10^{-5}$ ;  $n = 194$ ). None of the other polymorphisms analysed was significantly associated with the presence of disease.

**Conclusions:** Significant association was observed between dIBS and the SERT-P deletion/deletion genotype, suggesting that the serotonin transporter is a potential candidate gene for dIBS in women.

▲ *Gut* 2004;53:1452-1458.