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Oesophagus

ORIGINAL ARTICLE

Genetic variation in the *lymphotoxin- α (LTA)/tumour necrosis factor- α (TNF α)* locus as a risk factor for idiopathic achalasia

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ABSTRACT

Background Idiopathic achalasia is a rare motor disorder of the oesophagus characterised by neuronal loss at the lower oesophageal sphincter. Achalasia is generally accepted as a multifactorial disorder with various genetic and environmental factors being risk-associated. Since genetic factors predisposing to achalasia have been poorly documented, we assessed whether single nucleotide polymorphisms (SNPs) in genes mediating immune response and neuronal function contribute to achalasia susceptibility. **Methods** 391 SNPs covering 190 immune and 67 neuronal genes were genotyped in an exploratory cohort from Central European (CE) achalasia patients (n=704).

Significance of this study

What is already known on this subject?

- Achalasia is hypothesised to be an (auto) immune-mediated disease, possibly triggered by a viral infection such as herpes simplex virus 1, characterised by neuronal loss.
- Achalasia is a complex disorder with various genetic and environmental factors contributing to disease susceptibility.
- Former genetic studies suggested potential associations between achalasia and immune

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Rate of Detection of Advanced Neoplasms in Proximal Colon by Simulated Sigmoidoscopy vs Fecal Immunochemical Tests

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