Author's response to reviews

Title: Prevalence of Genetic Polymorphisms in the Promoter Region of the Alpha-1 Antitrypsin (SERPINA1) Gene in Chronic Liver Disease: a Case Control Study

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Please, find attached our manuscript entitled “Prevalence of genetic polymorphisms in the promoter region of the alpha-1 antitrypsin (SERPINA1) gene in chronic liver disease: a case control study” intended for publication in “BMC Gastroenterology”.

The c.-1973T>C mutation in the promoter region of the SERPINA1 gene appeared to be (partly) responsible for the pathogenesis of liver expression in Alpha-1 Antitrypsin Deficiency. We performed a genetic association study and reported the prevalence of c.-1973T>C in 297 patients with liver disease of various aetiologies and 297 controls.

We think that our study is of interest for the broad readership of BMC Gastroenterology because of the following reasons:

1. We showed that the prevalence of the c.-1973T>C polymorphism is not increased in patients with various liver diseases compared with healthy controls.
2. Our study was sufficiently powered to negate a 10% difference in polymorphism rate.
3. We showed an enrichment of S allele heterozygosity in patients with drug induced liver injury.

We take full responsibility for the integrity of the work as a whole. Each author has participated sufficiently in the intellectual content of the work to take public responsibility. None of its contents have been previously published, nor is it under consideration elsewhere. The authors do not have any financial interest. The manuscript represents original work and has been seen and approved by all authors. The authors do not have any commercial considerations for any product or process mentioned in the submission. All authors state that they have participated in the study and concur with the submission by the corresponding author. We would prefer to be contacted by e-mail (K.Kok@mdl.umcn.nl).

We sincerely hope that you are willing to consider our manuscript for publication and are looking forward to your response.

Yours sincerely,

Karin F. Kok and Joost P.H. Drenth