# Supplementary File 2. Genotype-phenotype correlation of ABCC2 variations.

Nonsense, frameshift, canonical splice sites, initiation codon loss, and single exon, or multiexon deletions were defined as null *ABCC2* variants; missense, synonymous, noncanonical splicing, and in-frame indels were defined as non-null ABCC2 variants. Based on the number of null or non-null ABCC2 variants, the patient genotypes were classified as biallelic-null, single-null, or biallelic non-null (Supplementary Table 2). The patients were also classified into two phenotypic groups by the presence or absence of acholic stools. Fisher’s exact test was performed using IBM SPSS version 19 (Armonk, NY, USA). *P*-values <0.05 were considered statistically significant. No significant correlations between acholic stool and genetic variation was observed (Supplementary Table 2). TB, DB, TBA, ALT, AST, GGT, and ALP levels were compared in patients with different genotypes. No significant correlations of different genotypes were observed for TBA and GGT levels (Supplementary Fig. 1).