Supplementary data to:

rs1944919 on chromosome 11q23.1 and its effector genes *COLCA1/COLCA2* confer susceptibility to primary biliary cholangitis

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Supplementary Tables

Table S1. Oligo-nucleotide probes for the EMSA

SNP	Allele	Sequence (5' -> 3')
rs4938534	T	TCCTCTTTCCCGACGTCGGGTAGTTTCTTAA
	C	TCCTCTTTCCCGACGCCGGGTAGTTTCTTAA
rs7952497	C	CCATCCCCCTTCCCCCGTTACAAAAAGGACA
	A	CCATCCCCCTTCCCCAGTTACAAAAAGGACA
rs6589227	T	TCCTGTGCGAAGTGGTTCGGTCTAGAGGTGA
	C	TCCTGTGCGAAGTGGCTCGGTCTAGAGGTGA
rs1944919	T	TCGAGGTGGGTACTGTCTTTCAACGGGTCCT
	G	TCGAGGTGGGTACTGGCTTTCAACGGGTCCT
rs6589226	G	TTCAAGTGTTAACAAGGTAGGTTGAGGCCCC
	A	TTCAAGTGTTAACAAAGTAGGTTGAGGCCCC
rs4356268	T	CCGATACGGGTGCCTTTTTCTCCGGTTGGGA
	C	CCGATACGGGTGCCTCTTTCTCCGGTTGGGA

Table S2. Primers for production of Luciferase assay constructs.

Primer Name	Sequence (5' -> 3')
rs1944919-F	GATATCTTTGCATGCATGCCATGCTC
rs1944919-R	AGATCTGAAAAACAGCAGTGCAGCTG
rs4938534-F	GGTACCTTCATGGTGTGCTTCACCTC
rs4938534-R	GAGCTCGCCTCGGGTATGTCTTTATC
rs7952497-F	GGTACCGCTTAAAACTGTGGAGGGAC
rs7952497-R	GAGCTCACATAGGAGCCCAATGAGAC
rs6589227-F	GGTACCCCTGTTTCTAGCTGAGTGAC
rs6589227-R	GAGCTCTTTCATCCAACTCCGGGGAT
rs6589226-F	GGTACCCTTCTGTGACAAGGCTTGTC
rs6589226-R	GAGCTCGGTTCAACTCTCAGAAGCTG
rs4356268-F	GGTACCGAGGCTCTGTTCATCTTGTG
rs4356268-R	GAGCTCTTGAAACACAGTAGCCCCTC

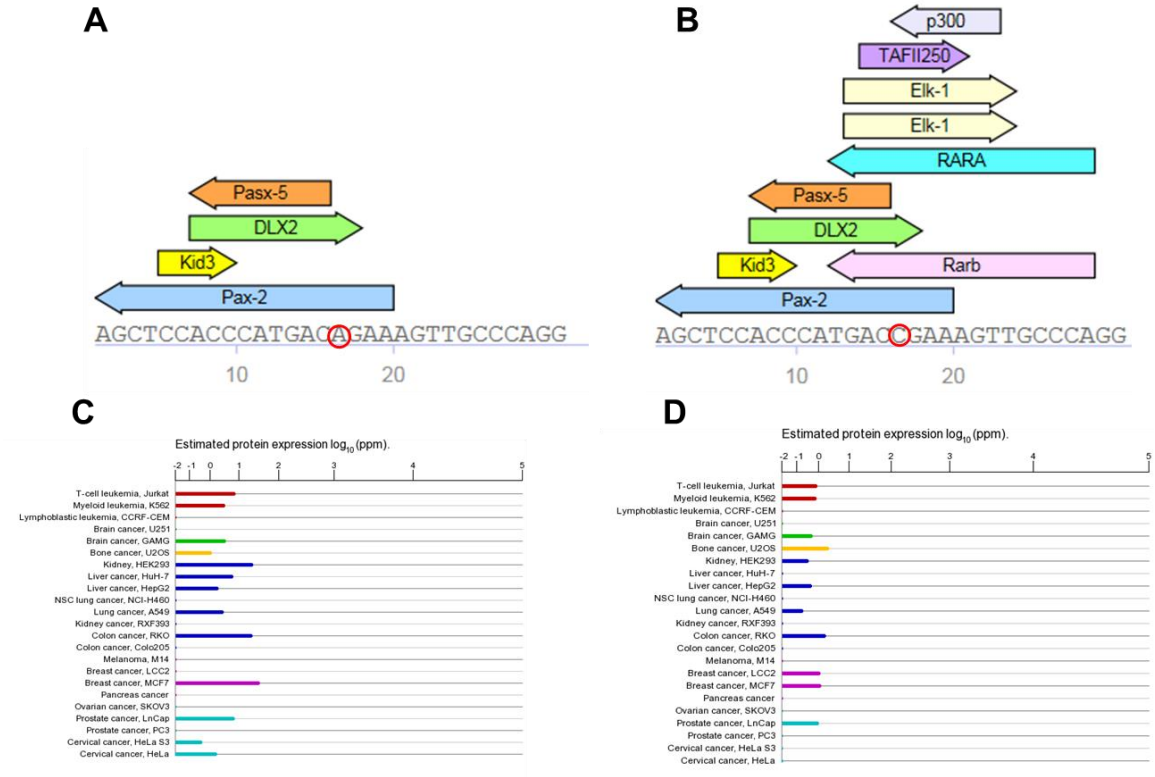
Table S3: Sequences of primers used for the synthesis of gRNAs, donor-DNA, and sequence check used for gene editing.

gRNA/donor DNA	Sequence
gRNA-1 primer-F	CCGGCCATGCTCCTGGGCAACTTT
gRNA-1 primer-R	AAACAAAGTTGCCCAGGAGCATGG
gRNA-2 primer-F	CCGGTCCTGGGCAACTTTCGGTCA
gRNA-2 primer-R	AAACTGACCGAAAGTTGCCCAGGA
gRNA-3 primer-F	CCGGTCCTGGGCAACTTTCTGTCA
gRNA-3 primer-R	AAACTGACAGAAAGTTGCCCAGGA
donor ssDNA-G	AACAGCAGTGCAGCTGCCAGCCCTATAGCT CCACACATGACCGAAAGTTGCCCAGGAGCA TGGCATGCATGCAAATAGACT
donor ssDNA-T	AACAGCAGTGCAGCTGCCAGCCCTATAGCT CCACACATGACAGAAAGTTGCCCAGGAGCA TGGCATGCATGCAAATAGACT
Sequence check-F	CAGGAGTTTCACCAGGTGTA
Sequence check-R	GAAAAACAGCAGTGCAGCTG

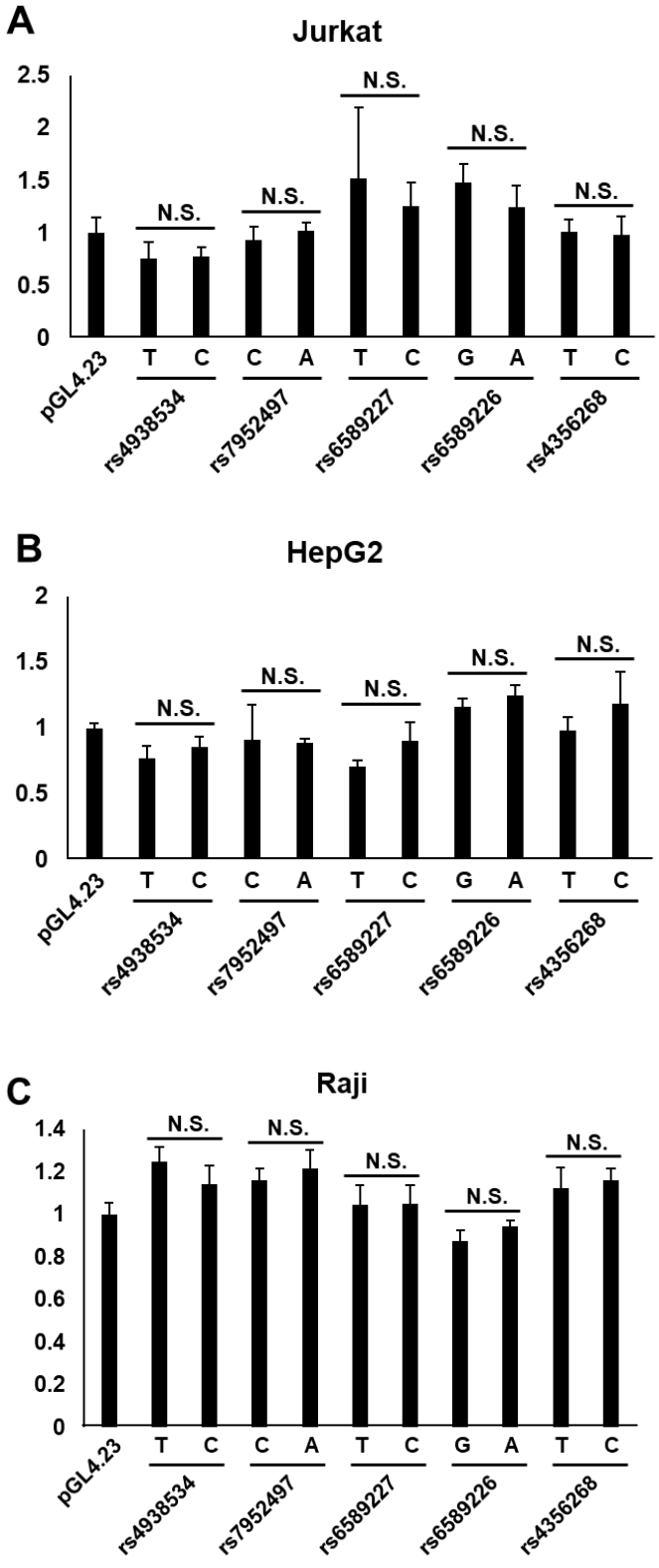
Table S4: Sequences of primers used for quantitative RT-PCR.

Gene Function	Gene name	amplicon size	Primer name	Sequence
Candidate effector gene	*POU2AF1*	71 bp	POU2AF1-F	TGGCGACCTACACCACAGT
			POU2AF1-R	CTCCTCTGTCACTGCAGAC
	COLCA1	155 bp	COLCA1-F	ATGTGGAGATGGACAGGGAT
			COLCA1-R	TCCTTGCTGTCCTTACAGAC
	COLCA2	113 bp	COLCA2-F	TCCGAGTGAAGATCACAGTG
			COLCA2-R	TGCAACTGGGTCTGAAAGGT
Housekeeping	*GAPHD*	194 bp	GAPDH-F	GCCAAGGTCATCCATGACAA
			GAPDH-R	TTCAGCTCAGGGATGACCTT

Supplementary Figures

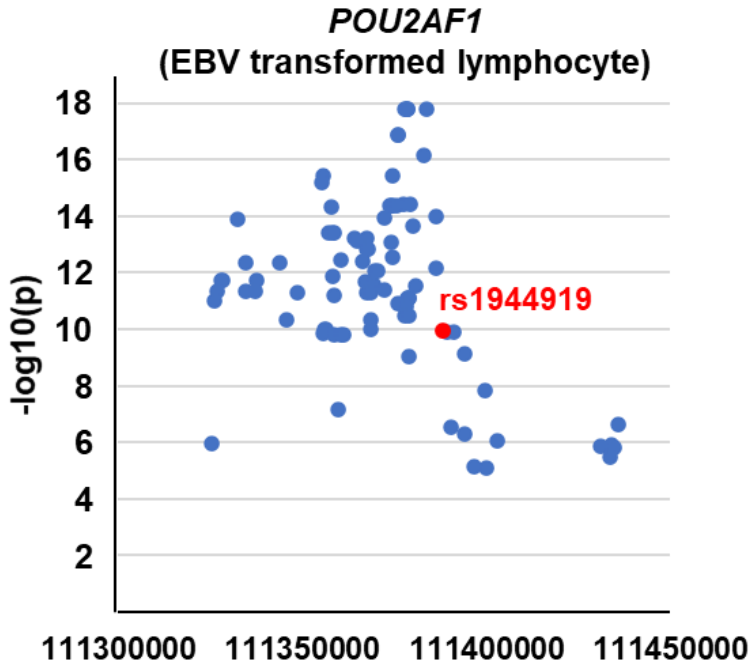


Supplementary Fig. 1. Prediction of transcription factor binding using the TRANSFAC database. (A) Predicted transcription factor binding in T allele of rs1944919. Reverse complement DNA sequences are shown. **(B)** Predicted transcription factor binding in G allele of rs1944919. Several transcription factors bind to G allele compared with T allele **(B)**. **(C and D)** EP300 **(C)** and TAF2 **(D)** expression levels in cancer cell lines. Both Jurkat and HepG2 cells showed abundant EP300 and TAF2 expression. Expression data were obtained using GeneCards from the Weizmann Institute of Science (http://www.genecards.org/).



Supplementary Fig. 2. Luciferase assay for other candidate SNPs.

Transcription was measured by cellular luciferase activity 24 h after transfection of Jurkat **(A),** HepG2 **(B),** and Raji **(C)** cells. No differences in luciferase activity were observed for the other candidate SNPs. Three independent experiments with triplicate measurements were performed for each assay. Data represent the mean ± SD; “N.S.” means non-significant (Student’s *t* test).



Supplementary Fig. 3. s-QTL mapping of *POU2AF1* in the EBV transformed lymphocyte. SNPs whose P-values less than 1.0×10^{-4} were shown. rs1944919, shown as red solid dots, did not exhibit the strongest association with *POU2AF1* splicing. rs4938534, which was the most significant SNP with PBC susceptibility on chromosome 11q23.1, did not associated with *POU2AF1* splicing ($P > 1.0 \times 10^{-4}$)