Supplementary Table 1. Characteristics of Individual Patients

No	Sex	Age at symptom onset, yr	Age at diagnosis, yr	Family history of CEAS	History of taking NSAIDs	Presenting symptoms	History of bowel resection	Involved sites	PHO manifestations	Tuberculosis tests*	Laboratory data						Genotype data of *SLCO2A1* gene	
											Hb, g/dL	Serum protein, g/dL	Serum albumin, g/dL	ESR, mm/hr	Serum CRP, mg/dL	Fecal calprotectin, μg/g	c.940+1G>A, splicing site (rs765249238)	c.1807C>T, p.R603X (rs776813259)
1	F	21	39	No	No	Abdominal pain, loose stool, weight loss	No	Jejunum, ileum except for terminal ileum	Digital clubbing, periostosis	Negative	7.9	4.9	1.9	2	1.11	10,679	Homozygous mutation	Wild type
2	F	32	45	No	No	Abdominal pain, weight loss	Yes	Ileum	Digital clubbing, arthralgia	NA	9.1	5.4	2.7	10	0.46	1,085	Heterozygous mutation	Heterozygous mutation
3	F	37	48	Yes	No	Abdominal pain	No	Stomach, duodenum, ileum, colon	NA	Negative	12.7	6.2	3.0	4	0.17	350	Wild type	Homozygous mutation
4	F	34	44	Yes	Yes	Abdominal pain	No	Stomach, duodenum, ileum	None	Negative	11.0	4.7	2.0	7	0.36	593	Wild type	Homozygous mutation
5	F	2	19	Yes	No	Abdominal pain	Yes	Ileum except for terminal ileum	NA	NA	7.3	4.6	2.3	3	0.14	4,765	Heterozygous mutation	Heterozygous mutation
6	M	11	54	Yes	No	Abdominal pain, loose stool, GI bleeding	Yes	Duodenum, jejunum, ileum, colon	Digital clubbing, pachydermia, periostosis	Negative	10.0	4.5	1.9	3	0.84	1,297	Heterozygous mutation	Heterozygous mutation
7	F	12	16	Yes	Yes	Abdominal pain, loose stool	No	Ileum	Arthralgia	NA	9.5	7.9	4.0	8	0.1	98.7	Heterozygous mutation	Heterozygous mutation
8	F	11	34	NA	No	Abdominal pain, loose stool, GI bleeding	Yes	Jejunum, ileum	NA	Negative	6.8	4.1	1.8	NA	NA	NA	Heterozygous mutation	Heterozygous mutation
9	F	12	21	No	No	Abdominal pain	No	Ileum	None	Negative	6.8	5.5	2.8	7	0.66	789	Heterozygous mutation	Heterozygous mutation
10	F	28	52	NA	Yes	Abdominal pain, loose stool	No	Ileum	Digital clubbing, pachydermia, periostosis	Negative	10.0	5.2	2.9	10	0.20	1,905	Wild type	Homozygous mutation
11	F	40	51	No	Yes	Abdominal pain	Yes	Jejunum, ileum except for terminal ileum	Arthralgia	Positive for IGRA	11.1	7.9	3.9	7	0.13	31.2	Heterozygous mutation	Heterozygous mutation

Supplementary Table 1. Continued

No	Sex	Age at symptom onset, yr	Age at diagnosis, yr	Family history of CEAS	History of taking NSAIDs	Presenting symptoms	History of bowel resection	Involved sites	PHO manifestations	Tuberculosis tests*	Laboratory data						Genotype data of *SLCO2A1* gene	
											Hb, g/dL	Serum protein, g/dL	Serum albumin, g/dL	ESR, mm/hr	Serum CRP, mg/dL	Fecal calprotectin, μg/g	c.940+1G>A, splicing site (rs765249238)	c.1807C>T, p.R603X (rs776813259)
12	F	NA	63	No	NA	Abdominal pain, loose stool, weight loss	No	Duodenum, jejunum, ileum	None	Negative	9.9	3.5	2.3	2	9.80	1,000	Homozygous mutation	Wild type
13	M	15	29	NA	No	Abdominal pain, loose stool	Yes	Duodenum, jejunum	Digital clubbing, pachydermia, periostosis	NA	8.2	5.3	2.7	3	0.12	211	Heterozygous mutation	Heterozygous mutation
14	F	NA	45	No	No	Abdominal pain, GI bleeding	Yes	Ileum	Arthralgia	Negative	9.7	6.2	4.3	11	7.72	152	Wild type	Homozygous mutation

CEAS, chronic enteropathy associated with *SLCO2A1* gene; NSAIDs, nonsteroidal anti-inflammatory drugs; PHO, primary hypertrophic osteoarthropathy; Hb, hemoglobin; ESR, erythrocyte sedimentation rate; CRP, C-reactive protein; F, female; M, male; GI, gastrointestinal; NA, not available; IGRA, interferon-gamma release assay.
*Tuberculosis tests include tuberculin skin test, interferon-gamma release assay, tuberculosis-specific ELISPOT assay, and acid-fast bacilli stain, *Mycobacterium tuberculosis* culture and polymerase chain reaction of intestinal biopsy tissue.

Supplementary Table 2. Presumed Nonpathogenic *SLCO2A1* Polymorphisms in 14 Korean Patients Confirmed to Have Chronic Enteropathy Associated with *SLCO2A1* Gene

No.	Genomic position chr3 (hg19)	Nucleotide change	Amino acid change	dbSNP	Mutant allele frequency*	gnomAD frequency†	Polyphen2	MutationTaster
1	133,666,209	c.1186G>A	p.A396T	rs34550074	0.286	2.53E-01	Benign	Polymorphism
2	133,935,791	c.210G>A	p.S70S	rs10935090	0.393	5.55E-01		Polymorphism
3	133,670,073	c.840A>G	p.R280R	rs6767412	0.429	2.54E-01		Polymorphism
4	133,654,635	c.2071C>T	p.N599N	rs117837593	0.429	2.49E-02		Polymorphism

*Mutant allele frequency in our 14 patients; †Genome Aggregation Database (gnomAD) East Asian frequency (GRCh37).

Supplementary Table 3. Presumed Nonpathogenic *SLCO2A1* Polymorphisms in 32 Korean Patients Who Were Not Confirmed to Have Chronic Enteropathy Associated with *SLCO2A1* Gene

No.	Genomic position chr3 (hg19)	Nucleotide change	Amino acid change	dbSNP	Mutant allele frequency*	GenomAD frequency†	Polyphen2	MutationTaster	ClinVar
1	133,666,209	c.1186G>A	p.A396T	rs34550074	0.266	2.53E-01	Benign	Polymorphism	Not reported
2	133,935,791	c.210G>A	p.S70S	rs10935090	0.344	5.55E-01		Polymorphism	Not reported
3	133,670,073	c.840A>G	p.R280R	rs6767412	0.297	2.54E-01		Polymorphism	Not reported
4	133,654,635	c.2071C>T	p.N599N	rs117837593	0.063	2.49E-02		Polymorphism	Benign
5	133,666,104	c.1279_1290del	p.Glu427_Pro430del	rs1085307096	0.016	5.44E-05			Likely pathogenic§
6	133,657,306	c.1657A>G	p.Ile553Val	rs199895359	0.016	4.66E-03	Benign	Disease-causing‡	Likely benign

*Mutant allele frequency in 32 patients in this study; †Genome Aggregation Database (gnomAD) East Asian frequency (GRCh37); ‡Grantham scores: 29 (chemical dissimilarity was designated "conservative"); §In the condition of primary hypertrophic osteoarthropathy, autosomal recessive 2.