

Table S1 The clinical and genetic mutation features of those reported in literatures along with our cases

Patients	P1	P2	P3	P4	P5	P6	P7	P8	P9	P10	P11	P12	P13	P14	P15	P16	P17	P18	P19	P20	p21	P22	P23	P24	
Sex	Male	Male	Male	Male	Male	Male	Male	Male	Male	Male	Male	Male	Male	Male	Male	Male	Male	Male	Male	Male	Male	Male	Male	Male	
Age of onset	2 y	9 y	13 y	2 m	17 y	10 y	5 y	1 y	4 y	20 d	10 y	10 y	3 y	na	3 y	2 y	10 y	3.5 y	3 m	3 y	6 y	10 y	6 y	11.5 y	
Inheritance	de novo	Mother	Mother	Mother	Mother	Mother	Mother	Mother	Mother	Mother	Mother	NA	Mother	Mother	Mother	Mother	Mother	Mother	Mother	Mother	Mother	Mother	Mother	Mother	
Mutation	p.W251S (c.752G>C)	p.W251S (c.752G>C)	p.Ala339fs*32 (c.1015delG)	p.W231R (c.691T>C)	p.T187N (c.560C>A)	p.R234X (c.700C>T)	p.Q39X (c.115C>T)	p.H156lfs*105 (c.465Gdel)	p.S248F (c.743C>T)	p.R185X (c.553C>T)	p.Q341Rfs*30 (c.1022del)	p.R234X (c.700C>T)	p.H156lfs*105 (c.465del)	p.G148Vfs*113 (c.443delG)	p.W212C (c.636G>T)	Deletion exons 2-7	Deletion exons 2-7	p.R234X (c.700C>T)	p.Arg267Trp (c.799C>T)	c.248-7G>A	p.R234X (c.700C>T)	p.P226L (c.677C>T)	p.Ala279ValfsTer103(c.835_836insTATACTACCA GTATACCAAAGAGGCA TACTGG)	p.S30Lfs*6 (c.87delC)	
Location	Exon 7 ETS domin	Exon 7 ETS domin	Exon 8	Exon 7 ETS domin	Exon 6	Exon 7 ETS domin	Exon 3	Exon 5	Exon 7 ETS domin	Exon 6	Exon 8	Exon 7 ETS domain	Exon 5	Exon 5	Exon 7 ETS domin	/	/	Exon 7 ETS domain	Exon 7 ETS domin	/	Exon 7 ETS domain	Intron	Exon 8	Exon 3	
Mutation type	Missense	Missense	Frameshift	Missense	Missense	Nonsense	Nonsense	Frameshift	Missense	Nonsense	Frameshift	Nonsense	Frameshift	Frameshift	Missense	Deletion	Deletion	Nonsense	Missense	NA	Nonsense	Missense	Frameshift	Frameshift	
Family history	NA	NA	NA	NA	NA	NA	Mother had Behçet's disease	NA	NA	NA	Mother had recurrent oral ulcers	NA	Mother had recurrent oral ulcers	NA	NA	NA	NA	NA	-	-	NA	NA	NA	-	
Clinical manifestation																									
Oral ulcers	+	+	+	+	NA	+	+	+	-	+	+	+	+	+	-	+	+	-	+	+	+	+	+	+	
Abdominal pain	+	-	-	-	NA	+	-	+	-	-	+	+	+	+	+	+	-	-	+	+	+	+	-	+	
Diarrhea	+	-	-	-	NA	-	-	+	-	+	+	-	+	-	+	+	-	+	-	-	+	-	-	-	
Intestinal ulcers	+	+	-	-	NA	+	-	+	-	+	+	+	+	NA	+	-	NA	+	+	+	+	+	+	+	
Perianal ulcers	-	+	-	-	NA	+	-	+	-	-	-	+	-	NA	NA	NA	NA	NA	-	-	-	-	+	-	
Perianal abscess	-	+	-	-	NA	-	-	+	-	-	-	-	+	NA	NA	NA	NA	NA	-	-	-	-	+	-	
Fever	+	+	+	+	NA	+	+	+	-	+	+	-	+	+	NA	+	-	+	+	+	+	+	+	-	
Arthritis	-	-	-	+	NA	-	+	-	-	-	-	-	+	-	-	-	-	-	-	-	-	-	-	-	
Recurrent infection	-	-	+	+	+	-	+	-	-	+	-	-	+	+	-	-	-	-	+	-	+	+	+	-	
Anemia	+	+	-	+	NA	+	+	+	-	+	-	-	-	+	NA	+	+	+	-	-	-	-	-	+	
Others	-	Perianal fistula	Skin inflammation	Constipation; palm rash	-	-	Skin erythema	-	-	Hydrocephalus, congenital hypospadias, inguinal and umbilical hernias	Erythema nodosum-like lesions	-	Rash	Widespread erythematous and non-pruritic micropapules	NA	Red tender papulonodular lesions with some overlying scale	Lesions on arms and legs are initially pustular and then drain and ulcerate	Skin inflammation described as bullous and prurigo-like lesions	Gastrointestinal perforation	Intestinal fistula; right hemicolectomy	Erythema nodosum-like lesions	-	-	Anal fistula	
Endoscopy																									
EGD	Gastritis	Gastritis, esophagitis	NA	NA	NA	NA	NA	Esophagus and gastric antral ulcers	NA	Gastric antral ulcers	NA	NA	The lower end of esophagus and the small flexure had shallow ulcers	NA	Esophagitis	Esophagitis	NA	Aphthous lesions in the esophagus and gastritis	NA	NA	Pharyngeal ulcer	-	Superficial gastritis	Superficial gastritis	
Colonoscopy	Aphthous-like ulcerations throughout colon	A single ulcerated lesion in the terminal ileum	NA	NA	NA	Terminal ileum, ileocecal flap and rectum isolated ulcers	NA	Multiple ulcers in the colon, stenosis of the ascending colon	NA	Multiple large circular ulcers in the ileal mucosa	Multiple ulcers in the ileocecal region	Terminal ileum, ileocecum, transverse colon, descending colon and rectum had irregular-shaped ulcers	The ascending colon was narrow, and irregular shape ulcers be found from ascending colon to rectum	NA	Continuous ulceration from the rectum through the cecu	Normal	NA	Terminal ileitis and pancolitis with erythematous and friable mucosa and multiple aphthous lesions	NA	Multiple irregular deep chisel ulcers at the anastomosis and terminal ileum	Terminal ileum and rectal ulcers	Massive ulceration in the terminal ileum	Multiple ulcers in the terminal ileum and colon	Large ileocecal valve ulcer	
Capsule endoscopy	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	terminal ileum ulceration	-	
Immunoglobulins	NA	Normal	NA	NA	IgG decreased	Normal	IgA increased	Normal	Normal	IgG increased	Normal	Normal	NA	IgG increased	NA	IgM and IgE increased	IgM and IgE increased	Normal	NA	NA	NA	IgA increased	Normal	IgA and IgG increased	
Immune cells	Intermittently low NK cells and CD27+ memory B cells	NA	NA	NK cells and memory B cells decreased	NK cells decreased	Normal	NK cells and B cells decreased	Normal	Intermittently low NK cells and B cells	Intermittently low NK cells and memory B cells	CD19 ⁺ B cells increased, NK cells was normal	Normal	Normal	NK cells decreased	NA	NK cells decreased	Normal	NK cells and memory B cells decreased	NK cells and B cells decreased	NA	NA	Intermittently low NK cells	Normal	NA	
Cytokines	NA	NA	NA	NA	NA	Normal	Normal	IL-1β, IL-8 and TNF-α increased	IL-6, IL-8, IL- 10, TNF-α and IFN-γ increased	Normal	IL-6 and IFN-γ increased	Normal	Normal	IL-6 and IL-18 increased	NA	IL-6 increased	Normal	IL-6, IL-10 and IL-18 increased	NA	NA	NA	NA	Normal	NA	
Treatment																									
Mesalazine	-	-	-	-	NA	+	-	-	-	-	-	+	-	-	-	-	-	-	-	-	+	+	-	-	
Glucocorticoid	+	+	+	+(topical)	NA	+	+	+	+	+	+	+	+	+	+	+	+	+	+	-	+	+	-	+	
IVIg	-	-	-	+	NA	-	-	-	-	+	-	-	-	-	-	-	-	-	-	-	-	-	-	-	
EN	-	-	-	-	NA	-	-	-	-	-	-	-	+	-	-	-	-	-	-	-	-	-	+	-	
Biologics	Canakinumab	Ustekinumab	Adalimumab	-	Na	-	Infliximab	-	Benlysta	Infliximab	Adalimumab	-	-	-	Infliximab	Adalimumab, canakinumab	Adalimumab	Infliximab	Infliximab; adalimumab; thalidomide; tocilizumab	Adalimumab	Infliximab	Adalimumab	Adalimumab	-	
NSAIDS	-	-	-	+	NA	-	+	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	
Mercaptopurine/ Thiopurine	-	-	-	-	NA	-	-	-	-	-	-	-	+	-	+	-	-	+	-	-	-	-	-	+	
Others	Anakinra	-	-	Methotrexate	NA	-	-	-	-	-	Thalidomide	-	-	-	Methotrexate	Colchicine, Apremilast	-	-	ciclosporin	Thalidomide	-	Thalidomide	-	-	
Response	remission	remission	remission	remission	NA	remission	remission	remission	remission	remission	remission	remission	remission	remission	NA	remission	remission	remission	partial remission	partial remission	remission	remission	remission	remission	
Reference	(7)	(7)	(7)	(8)	(9)	(2)	(2)	(2)	(2)	(2)	(13)	(10)	(10)	(14)	(14)	(14)	(14)	(14)	(11)	(11)	(12)	(12)	This paper	This paper	