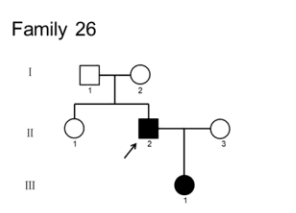
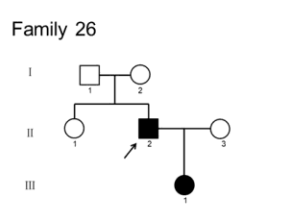
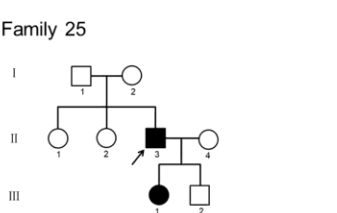
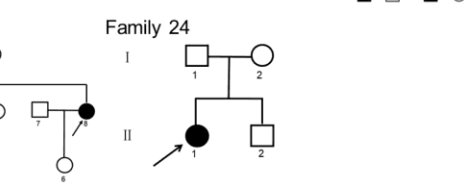
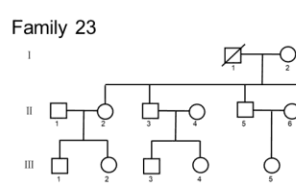
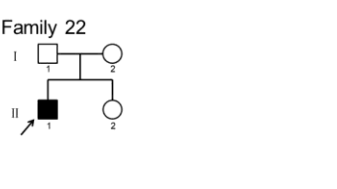
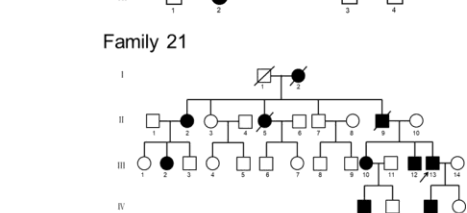
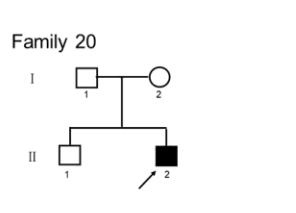
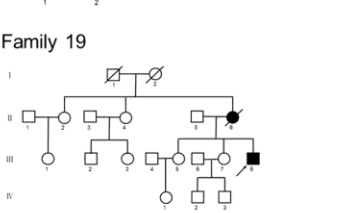
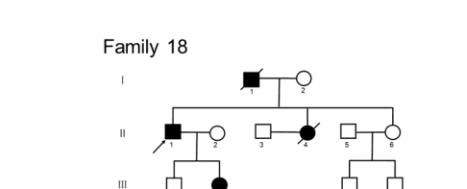
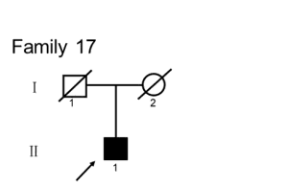
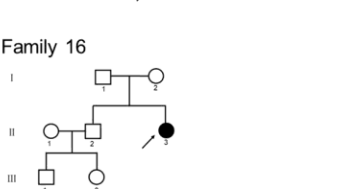
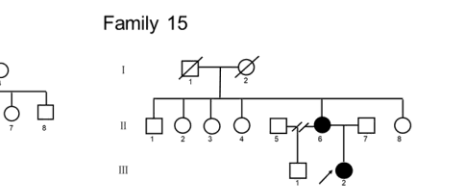
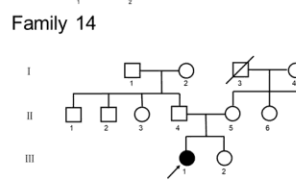
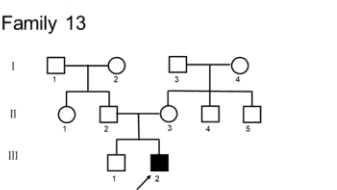
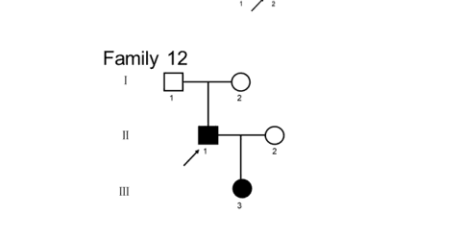
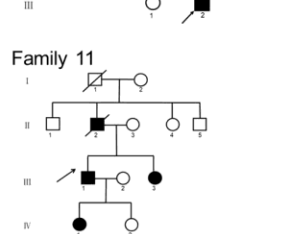
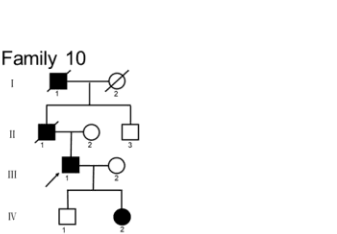
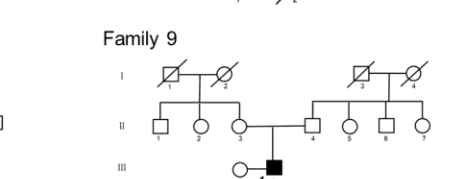
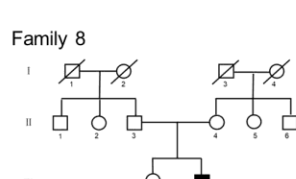
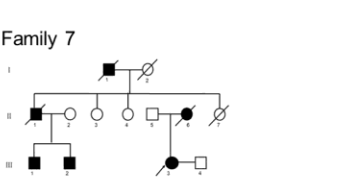
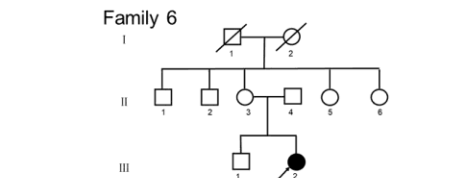
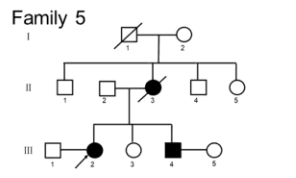
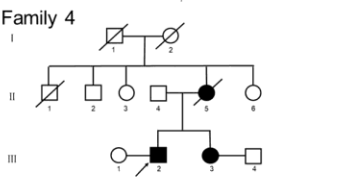
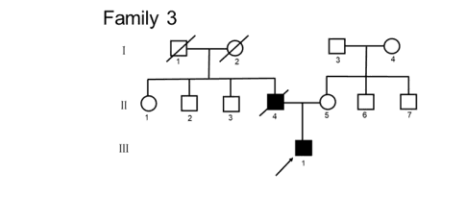
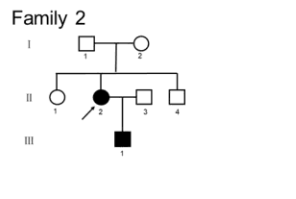
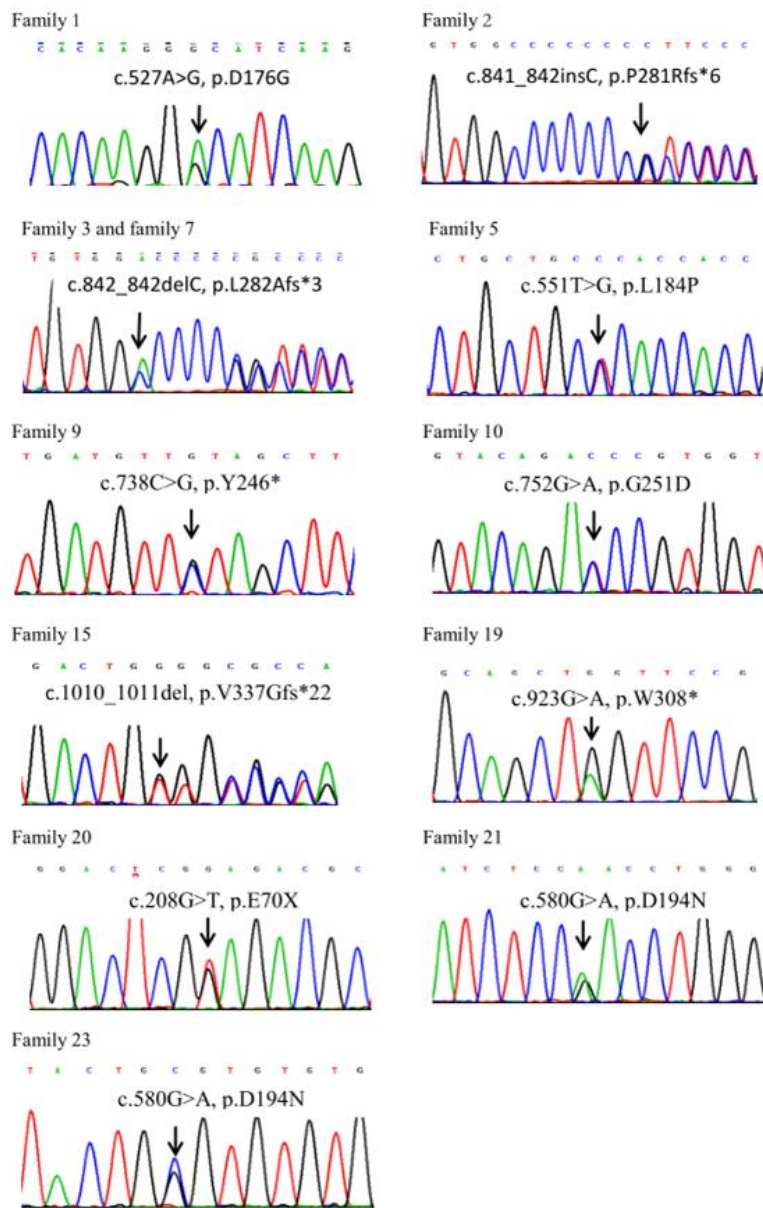


Family 1



Supplementary Figure 1: The pedigree of 26 families. Family members are identified by generations and numbers. Circles and squares denote females and males, respectively; blackened symbols indicate the PJS patient; the proband is indicated by an arrow. Symbols with a slash indicate the individual was already dead.



Supplementary Figure 2: Chromatogram of 11 other *STK11* gene mutations;

mutations are indicated with arrows.

Table 1 Primers of *STK11* gene

	Forward primer	Reverse primer
Exon 1	5'-gggactgacgtgtagaacaatc-3'	5'-ggaaggaagacagaacccatcag-3'
Exon 2	5'-tctaggggaaggaggaggta-3'	5'-tcaaggagacgggaagagg-3'
Exon 3	5'-cctgagctgtgtgtccttag-3'	5'-gggccttcattgtcaatgaatc-3'
Exon 4-5	5'-ccttcctctgtcctgtgtg-3'	5'-ccatctgccgtatgagttacat-3'
Exon 6	5'-actgaccacgccttcctc-3'	5'-tctccactcagtcctcctcaat-3'
Exon 7	5'-ttaggagcgtccaggtatca-3'	5'-gccagtgttgggaccag-3'
Exon 8	5'-catggctgagcttctgtgg-3'	5'-gaggccttggggacgtgggattgg-3'
Exon 9	5'-gggggcagcatttcagg-3'	5'-ggcatccaggcgttgc-3'

Supplementary table 2 Detailed clinical and genetic data for patients with PJS

Supplementary table 2 Detailed clinical and genetic data for patients with type 1														
Family		Mutation		Patient			Initial evaluation		First detection of polyps		Clinical course***			
ID	Exon	Nucleotide	Effect	ID	Sex*	Current Age(y)	Age(y)	Main Reason*	Age(y)	Main Reason**	AP	GB	IB	Surgery
1	4	c.527A>G	p.D176G	1	M	26	20	MP	20	MP	Y	Y	Y	Y
				2	M	57	53	FH	—	—	N	N	N	N
				3	M	27	23	IB	15	IB	Y	Y	Y	Y
				4	F	9	5	FH	—	—	N	N	N	N
				5	M	2	0	FH	—	—	N	N	N	N
2	6	c.842_842insC	p.L282Afs*3	6	F	42	16	GB	16	GB	Y	Y	Y	Y
				7	M	18	14	GB	14	GB	Y	Y	N	N
3	6	c.841_842delC	p.P281Rfs*6	8	M	24	17	GB	17	GB	Y	Y	N	Y
4	7	c.889A>G	p.R297G	9	M	34	22	GB	22	GB	N	Y	N	Y
				10	F	31	25	FH	25	FH	Y	Y	N	N
5	4	c.551T>C	p.L184P	11	F	40	21	FH	21	FH	N	N	N	N
				12	M	35	20	FH	20	FH	N	N	N	N
6		Het del exon 1		13	F	32	23	IB	23	IB	Y	N	Y	Y
7	6	c.841_842insC	p.L282Afs*3	14	F	31	16	IB	16	IB	Y	Y	Y	Y
8		Het del exon 1		15	M	18	11	IB	11	IB	Y	Y	Y	Y
9	6	c.738C>G	p.Y246*	16	M	40	15	IB	15	IB	Y	N	Y	Y
10	6	c.752G>A	p.G251D	17	M	37	29	GB	29	GB	N	Y	N	N
11		Het del exon 1		18	M	35	26	IB	26	IB	Y	N	Y	Y
				19	F	33	20	FH	20	FH	Y	N	N	N
				20	M	45	20	IB	20	IB	Y	N	Y	Y
12	3	c.457_458insCC	p.H154Pfs*8	21	F	21	13	IB	13	IB	Y	N	Y	Y
13		Het del exon 2-3		22	M	9	2	GB	5	GB	Y	Y	N	N
14	3	c.393C>A	p.Y131*	23	F	22	15	IB	15	IB	Y	Y	Y	Y
15	8	c.1010_1011delTG	p.V337Gfs*22	24	F	21	17	GB	17	GB	N	Y	N	N
16		Het del exon 8		25	F	8	5	SV	5	SV	Y	N	N	Y
17	3	c.428_428delT	p.V143Gfs*18	26	M	19	2	GB	2	GB	Y	Y	Y	Y
18		Het del exon 2-3		27	M	50	43	GB	43	GB	Y	Y	N	N
19	8	c.923G>A	p.W308*	28	M	31	22	GB	22	GB	Y	Y	N	N
20	1	c.208G>T	p.E70*	29	M	18	11	IB	11	IB	Y	N	Y	Y
21	4	c.580G>A	p.D194N	30	M	32	14	IB	14	IB	Y	N	Y	Y
				31	M	28	27	FH	27	FH	N	N	N	N
22	5	c.601_601delC	L201Cfs*86	32	M	23	22	IB	22	IB	Y	N	Y	Y
23	3	c.396C>G	C132W	33	F	50	20	IB	20	IB	Y	Y	Y	Y
24	8	c.930delG	p. K311Rfs*25	34	F	7	6	MP	—	—	N	N	N	N
25	5	c.716G>C	p.W239S	35	M	34	20	D	20	D	Y	N	N	N
				36	F	9	9	FH	—	—	N	N	N	N
26	2	c.358G>T	p. E120*	37	M	31	10	MP	18	MP	Y	Y	Y	N
				38	F	4	4	FH	—	—	N	N	N	N

*M=male; F=female; **MP=mucocutaneous hyperpigmentation; FH=family history; GB=gastrointestinal bleeding; IB=intestinal obstruction; SV=Stomach Volvulus; D=diarrhea; "—"represents did not do any intestinal-associated testing; ***AP=abdominal pain; Y=yes; N=no