

**Supplementary Table 1 Summary of family members of patients with Peutz-Jeghers syndrome**

Family No.	Proband gender	PJS family history	Family members					
			Relationship with proband	Age (y)	STK11 gene pedigree analysis	Medical history	Pigmentation	Polyp
1	Man	<i>De novo</i>	/		/	/	/	/
2	Woman	Yes, maternal. Mother, brother.	Mother	44	Died	Died of cervical cancer at 44y.	Yes, lips and fingers	No examination
			Father	33	None	None	None	No examination
			Brother	56	Yes, two variants	Sinonasal cancer diagnosed at 26y.	Yes, lips, oral, fingers, and toes	Yes, colon
			Older aunt	63	Yes, one variation c.1062C>G	None	None	None
3	Man	<i>De novo</i>	Fahter	55	None	None	None	None
			Mother	50	None	None	None	None
4	Man	<i>De novo</i>	Fahter	56	None	None	None	None
			Mother	54	None	None	None	None
5	Woman	Yes, paternal. Father, younger uncle, brother.	Fahter	61	undetected			
			Mother	59	None	None	None	None
			Brother	30	undetected	PJS diagnosed at 7y.	Yes, lips, oral, fingers	Yes, gastrointestinal tract

			Son	2	None	None	None	None
			Older uncle	65	None	None	None	None
			Younger uncle	19	Died	Died of PJS at 19y.	Yes	Yes, gastrointestinal tract
6	Woman	<i>De novo</i>	Fahter	57	None	None	None	None
			Mother	57	None	None	None	None
7	Woman	<i>De novo</i>	Fahter	79	None	None	None	None
			Mother	73	None	None	None	None
8	Man	Yes, paternal. Father, grandfather, sister and niece.	Father	58	Yes	None	Yes, lips, cheek, fingers, toes	No examination
			Grandfather	85	Died	Stroke at 60y, cerebrovascular disease at 81y.	Yes, lips, fingers	No examination
			Sister	28	undetected	None	Yes, lips, cheek, toes	No examination
			Older niece	3	undetected	None	Yes, oral	No examination
			Younger niece	1	undetected	None	None	No examination
9	Woman	<i>De novo</i>	Fahter	57	None	None	None	None
			Mother	56	None	None	None	None

Abbreviation: y, years.