S2 Table. Types of mutation reported in previous studies of Korean patients with Li-Fraumeni syndrome

Study	Study design (patient number)	Nucleotide alteration	Amino acid alteration	Mutation type	Frequency
Park et al. (2016) [1]	Case series (14)	c.78delT	p.Pro27Leufs*17	Frameshift	1
		c.91G>A	p.Val31Ile	Missense	2
		c.293delC	p.Pro98Leufs*25	Frameshift	1
		c.524G>A[a)]	p.Arg175His[a)]	Missense	3
		c.566C>T	p.Ala189Val	Missense	1
		c.742C>T[a)]	p.Arg248Trp[a)]	Missense	1
		c.818G>A[a)]	p.Arg273His[a)]	Missense	3
		c.817C>T[a)]	p.Arg273Cys[a)]	Missense	1
		c.859G>T	p.Glu287*	Nonsense	1
Alyami et al. (2021) [2]	Case series (12)	c.78delT	p.Pro27Leufs*17	Frameshift	1
		c.375G>A	p.Thr125=	Silent	1
		c.524G>A[a)]	p.Arg175His[a)]	Missense	1
		c.542G>A	p.Arg181His	Missense	1
		c.566C>T	p.Ala189Val	Missense	1
		c.590T>G	p.Val197Gly	Missense	1
		c.638G>A	p.Arg213Gln	Missense	1
		c.730G>A	p.Gly244Ser	Missense	1
		c.743G>A	p.Arg248Gln	Missense	2
		c.742C>T[a)]	p.Arg248Trp[a)]	Missense	1
		c.824G>A	p.Cys275Tyr	Missense	1
Bang et al. (1995) [3]	Case report (1)	c.742C>T[a)]	p.Arg248Trp[a)]	Missense	1
Kim et al. (2005) [4]	Case report (1)	c.859G>T	p.Glu287*	Nonsense	1
Baek et al. (2019) [5]	Case report (1)	c.542G>A	p.Arg181His	Missense	1
Hwang et al. (2008) [6]	Case report (1)	c.524G>A[a)]	p.Arg175His[a)]	Missense	1
Shin et al. (2009) [7]	Case report (1)	c.818G>A[a)]	p.Arg273His[a)]	Missense	1
Cho et al. (2013) [8]	Case report (1)	c.566C>T	p.Ala189Val	Missense	1
Oh et al. (2014) [9]	Case report (1)	c.524G>A[a)]	p.Arg175His[a)]	Missense	1
Ku et al. (2020) [10]	Case report (1)	c.743G>A	p.Arg248Gln	Missense	1

[a)]The same mutations as in this study.

References

1. Park KJ, Choi HJ, Suh SP, Ki CS, Kim JW. Germline TP53 mutation and clinical characteristics of Korean patients with Li-Fraumeni syndrome. Ann Lab Med. 2016;36:463-8.
2. Alyami H, Yoo TK, Cheun JH, Lee HB, Jung SM, Ryu JM, et al. Clinical features of breast cancer in South Korean patients with germline TP53 gene mutations. J Breast Cancer. 2021;24:175-82.
3. Bang YJ, Kang SH, Kim TY, Jung CW, Oh SM, Choe KJ, et al. The first documentation of Li-Fraumeni syndrome in Korea. J Korean Med Sci. 1995;10:205-10.
4. Kim IJ, Kang HC, Shin Y, Yoo BC, Yang HK, Park JG. Familial gastric cancers with Li-Fraumeni syndrome: a case repast. World J Gastroenterol. 2005;11:4124-6.
5. Baek YS, Seo JY, Song JY, Lee SY, Kim A, Jeon J. Li-Fraumeni syndrome presenting as cutaneous melanoma in a child: case report and review of literature. J Eur Acad Dermatol Venereol. 2019;33:e174-5.
6. Hwang SM, Lee ES, Shin SH, Kong SY. Genetic counseling can influence the course of a suspected familial cancer syndrome patient: from a case of Li-Fraumeni like syndrome with a germline mutation in the TP53 gene. Korean J Lab Med. 2008;28:493-7.
7. Shin HJ, Kwon YJ, Lim YJ, Park BK, Ghim TT, Shin SH, et al. Family of Li-Fraumeni syndrome with a germline mutation in the p53 gene. Clin Pediatr Hematol Oncol. 2009;16:38-42.
8. Cho Y, Kim J, Kim Y, Jeong J, Lee KA. A case of late-onset Li-Fraumeni-like syndrome with unilateral breast cancer. Ann Lab Med. 2013;33:212-6.
9. Oh CS, Lee JH, Jung ST, Na BR. Osteosarcoma with adenocarcinoma of lung in Li-Fraumeni

syndrome: a case report. J Korean Bone Joint Tumor Soc. 2014;20:99-103.
10. Ku I, Park JU. Dermatofibrosarcoma in Li-Fraumeni syndrome with early-onset multiple primary tumors. J Dermatol. 2020;47:e333-5.