

SUPPLEMENTARY TABLE S1

Case ID	Centers (1)	Case Classification	Gene Affected	Sex	Suspected Syndrome	Family History Polyps	Family History CRC or other cancer	Polyps Type Classification	Polyps Count	Polyps Histology	Polyposis Age at diagnosis	1st Cancer (Age at diagnosis)	2nd Cancer (Age at Diagnosis)	Extracolonic Manifestations 1st type (Age at diagnosis)	Extracolonic Manifestations 2nd type (Age at diagnosis)
002	HGBU	Patient	APC	M	FAP	Yes	No	FAP	>1000	No	No	No	No	No	No
004	HGBU	Patient	APC	M	FAP	Yes	Mother Uterus	FAP	Severe (>1000)	Adenoma	33	CRC (33)	No	Desmoid Tumor (33)	Duodenal Adenomas
005	HGBU	Patient	APC	M	FAP	No	No	FAP	Severe (>1000)	FAP	24	CRC (24)	No	No	Duodenal Adenomas
010	HGBU	Patient	APC	F	FAP	No	No	FAP	Severe (>1000)	FAP	29	CRC (29)	No	No	Duodenal Adenomas
011	HGBU	Patient	APC	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	17	No	No	Desmoid Tumor	OSTEOMA
012	HGBU	Patient	APC	M	FAP	Yes	Uncle CRC	FAP	Severe (>1000)	FAP	15	No	No	No	No
013	HGBU	Patient	APC	M	FAP	Yes	Father Sigmoid	FAP	Severe (>1000)	FAP	34	No	No	No	No
014	HGBU	Patient	APC	M	FAP	No	No	FAP	Severe (>1000)	FAP	17	No	No	No	No
016	HGBU	Patient	APC	M	FAP	No	No	FAP	Severe (>1000)	FAP	30	CRC (30)	No	Cancer Duodenum	No
020	HGBU	Patient	APC	F	FAP	Yes	Mother CRC	FAP	Severe (>1000)	FAP	35	No	No	Duodenal Adenomas	No
023	HGBU	Patient	APC	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	35	No	No	Gastric Polyps (35)	Abdominal desmoid (36)
030	HGBU	Patient	APC	F	FAP	Yes	No	FAP	Severe (>1000)	Adenomas	26	No	No	Duodenal Polyps (29)	No
031	HGBU	Patient	APC	F	FAP	No	No	FAP	Severe (>1000)	Adenomas	21	No	No	No	No
032	HGBU	Patient	APC	F	FAP	Yes	Yes	FAP	Severe (>1000)	FAP	48	No	No	No	No
033	HGBU	Patient	APC	F	FAP	Yes	Father CRC	FAP	Severe (>1000)	FAP	14	No	No	No	No
034	HGBU	Patient	APC	M	FAP	Yes	Yes	FAP	Severe (>1000)	FAP	14	No	No	No	No
035	HGBU	Patient	APC	M	FAP	No	No	FAP	Severe (>1000)	FAP	27	No	No	Soft Tissue (3)	Desmoid (29)
040	HGBU	Patient	APC	M	FAP	No	No	FAP	Severe (>1000)	FAP	34	CRC (34)	No	No	No
042	HGBU	Patient	APC	F	FAP	No	Father CNS Cancer	AFAP	20	AFAP	56	No	No	No	No
043	HGBU	Patient	APC	F	FAP	No	Aunt Uterus and CRC; Cousin: Thyroid	FAP	Severe (>1000)	Adenomas	36	No	No	No	No
045	HGBU	Patient	APC	F	FAP	Yes	Mother, Sister: Rectum cancer	FAP	Severe (>1000)	Adenomas	17	No	No	Gastric Polyps (17)	No
047	HGBU	Patient	APC	M	FAP	Yes	Father CRC	Mixed/Attenuated	Severe	Serrated Adenomas	26	CRC (60)	No	No	No
050	HGBU	Patient	APC	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	22	No	No	LIPOMA (24)	OSTEOMA (24)
052	HGBU	Patient	APC	F	FAP	Yes	Sister CRC	FAP	Severe (>1000)	Adenomas	15	No	No	No	No
053	HGBU	Patient	APC	F	FAP	No	Mother CRC	FAP	Severe (>1000)	Adenomas	33	CRC (33)	No	No	No
055	HGBU	Patient	APC	M	FAP	Yes	CRC	FAP	Severe (>1000)	Adenomas	33	No	No	LIPOMA	No
057	HGBU	Patient	APC	F	FAP	Yes	No	AFAP / Mixed	<100	Hiperplastic Adenomas	30	No	No	No	No
058	HGBU	Patient	APC	F	FAP	Yes	Mother Rectum, Aunt Duodenum / Thyroid	FAP	Severe (>1000)	Adenomas	29	No	No	Gastric Polyps (30)	No
060	HGBU	Patient	APC	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	45	CRC (47)	No	Gastric Polyps (45)	No
069	HGBU	Patient	APC	F	FAP	No	No	AFAP	<100	Juvenile	22	No	No	No	No
071	HGBU	Patient	APC	F	FAP	Yes	Yes	AFAP	Severe (>1000)	Adenomas	24	No	No	No	No
075	HGBU	Patient	APC	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	19	CRC (21)	No	No	No
077	HGBU	Relative to case 033	APC	F	FAP	Yes	No	FAP	Severe (>1000)	Adenomas	30	No	No	No	No
079	HIBA	Patient	APC	M	FAP	Yes	No	FAP	Severe (>1000)	Adenomas	37	Sarcoma (37)	No	Thyroid Cancer (40)	No
080	HIBA	Patient	APC	F	FAP	Yes	No	FAP	Severe (>1000)	Adenomas	32	CRC (48)	CRC (32)	No	No
081	HIBA	Patient	APC	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	42	CRC (42)	No	Desmoid Tumors (34, 36)	Rectal and Anal Polyps (49)
082	HIBA	Patient	APC	M	FAP	No	No	FAP	Severe (>1000)	Adenomas	50	No	No	No	No
083	HIBA	Patient	APC	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	50	No	No	No	No
084	HIBA	Patient	APC	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenoma High Dysplasia	35	CRC (61)	No	No	No
085	HIBA	Patient	APC	M	FAP	Yes	Yes	FAP	Severe (>1000)	Hyperplastic Polyps; Adenoma Low Grade Dysplasia	31	No	No	Desmoid Tumor (35)	No
086	HIBA	Patient	APC	F	FAP	No	No	FAP	Severe (>1000)	Adenomas	37	No	No	Pancreatic (55)	No
087	HIBA	Patient	APC	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	37	No	No	No	No
088	HIBA	Patient	APC	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	24	No	No	No	No
089	HIBA	Patient	APC	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	15	No	No	No	No
090	HIBA	Patient	APC	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	36	No	No	No	No
091	HIBA	Patient	APC	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	17	No	No	Thyroid Cancer (35)	No
092	HIBA	Patient	APC	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	33	CRC (34)	Rectum Adenocarcinoma	No	No
093	HIBA	Patient	APC	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	20	No	No	No	No
094	HIBA	Patient	APC	M	FAP	Yes	Yes	AFAP	Severe (>1000)	Adenoma Low grade dysplasia	26	No	No	No	No
095	HIBA	Patient	APC	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenoma High grade Dysplasia	50	No	No	Gastric and Duodenal Polyps, low grade dysplasia (28)	Adenoma of the ampulla of Vater (52)
096	HIBA	Patient	APC	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenoma High grade Dysplasia	28	CRC (28)	No	Stomach Polyps (28)	No
097	HIBA	Patient	APC	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenoma Low grade dysplasia	12	No	No	No	No
098	HIBA	Patient	APC	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	52	No	No	No	No
099	HIBA	Patient	APC	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	29	No	No	No	No
100	HIBA	Patient	APC	F	FAP	No	No	FAP	Severe (>1000)	Adenomas	55	Breast Cancer (55)	CRC (75)	No	No
101	HIBA	Patient	APC	M	FAP	No	No	FAP	Severe (>1000)	Adenomas	38	CRC (38)	No	No	No
102	HIBA	Patient	APC	M	FAP	No	No	FAP	Severe (>1000)	Adenomas	38	CRC (38)	No	No	No
103	HIBA	Patient	APC	F	FAP	No	No	FAP	Severe (>1000)	Adenomas	30	No	No	No	No
104	HIBA	Patient	APC	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	30	No	No	No	No
113	HIBA	Relative to case 095	APC	M	FAP	No	No	FAP	Severe (>1000)	Adenomas	30	No	No	No	No
115	HIBA	Patient	APC	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	32	No	No	No	No
170	HEC	Patient	APC	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	18	No	No	Duodenal Adenomas (18)	No
171	HEC	Patient	APC	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	23	No	No	Duodenal Adenomas (30)	No
173	HEC	Patient	APC	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	15	No	No	Duodenal Adenomas (29)	Fundic Gland Polyps (26)
174	HEC	Patient	APC	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	12	No	No	Duodenal Adenomas (13)	Fundic Gland Polyps (13)
179	HEC	Patient	APC	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	23	CRC (23)	No	duodenal papilla adenocarcinoma (32)	Fundic Gland Polyps (32)
183	HEC	Patient	APC	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	12	No	No	No	No
185	HEC	Relative to case 170	APC	NA	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	No	No	No	No	No
186	HEC	Relative to case 170	APC	NA	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	No	No	No	No	No
187	HEC	Relative to case 174	APC	NA	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	No	No	No	No	No
188	HEC	Relative to case 174	APC	NA	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	No	No	No	No	No
048	HGBU	Patient	BMPR1A	M	FAP	No	No	FAP	Severe (>1000)	Adenomas	25	No	No	No	No
109	HIBA	Patient	BMPR1A	F	FAP	No	No	FAP	Severe (>1000)	Adenomas	48	CRC (48)	No	No	No
001	HGBU	Patient	BRCA1	M	FAP	No	No	FAP	Severe (>1000)	Adenomas	42	No	No	No	No
049	HGBU	Patient	ERCC2	F	FAP	No	Father Kidney	AFAP	30	Adenomas	52	No	No	No	No
003	HGBU	Patient	FANCM	M	FAP	No	Grandfather CRC	AFAP	20	Adenoma	58	CRC (58)	No	No	No
029	HGBU	Patient	FLCN	M	FAP	No	No	AFAP	<100	Adenomas	64	No	No	No	No
110	HIBA	Patient	MSH3	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	42	No	No	No	No
048	HGBU	Patient	MSHC	M	FAP	No	Uncle Pancreas, Aunt: Ovary, Breast	AFAP	20	Adenomas	57	CRC (57)	No	No	No
017	HGBU	Patient	MUTYH	F	FAP	No	No	FAP	>100	FAP	43	CRC (43)	No	Thyroid Nodule (43)	No
024	HGBU	Patient	MUTYH	F	FAP	No	No	FAP	>100	FAP	37	CRC (37)	No	No	No
078	HIBA	Patient	MUTYH	M	FAP	Yes	No	FAP	>100	Adenomas	37	CRC (72)	No	No	No
105	HIBA	Patient	MUTYH	M	FAP	Yes	Yes	FAP	>100	Adenomas	50	No	No	No	No
106	HIBA	Patient	MUTYH	F	FAP	No	No	FAP	>100	Adenomas	37	CRC (63)	No	No	No
107	HIBA	Patient	MUTYH	M	FAP	No	No	FAP	>100	Adenomas	37	CRC (47)	No	No	No
108	HIBA	Patient	MUTYH	M	FAP	Yes	Yes	FAP	>100	Adenomas	57	No	No	No	No
111	HIBA	Relative to case 105	MUTYH	F	FAP	No	No	FAP	>100	Adenomas	No	No	No	No	No
112	HIBA	Relative to case 106	MUTYH	F	FAP	Yes	Yes	FAP	>100	Adenomas	38	No	No	No	No
114	HIBA	Patient	MUTYH	F	FAP	Yes	Yes	FAP	>100	Adenomas	45	No	No	No	No
046	HGBU	Patient	NEIL1	M	FAP	No	Yes (Father CRC)	AFAP / Mixed	>100	Serrated Adenomas	60	CRC (60)	No	No	No
006	HGBU	Patient	SMAD4	F	FAP	No	No	AFAP	20	Mixed	60	No	No	Gastric Polyps	No
027	HGBU	Patient	SMAD4	M	FAP	No	Yes	AFAP	<100	Juvenile	18	No	No	No	No
007	HGBU	Patient		M	FAP	Yes	Father CRC	FAP	Severe (>1000)	FAP	26	No	No	No	No

008	HGBU	Patient	F	FAP	No	No	FAP	>100	Adenomas	55	CRC (55)	Thyroid Nodule	
009	HGBU	Patient	M	FAP	Yes	No	AFAP	<100	Adenomas	51	CRC	Osteoma	Thyroid Nodules
015	HGBU	Patient	F	FAP	Yes	Yes	FAP	>100	FAP	10	No	Duodenal Adenomas	
018	HGBU	Patient	M	FAP	No	Father Prostate	FAP	Severe (>1000)	FAP	65	No	No	
021	HGBU	Patient	F	FAP	No	No	FAP	<100	Adenomas	27	No	No	
022	HGBU	Patient	M	FAP	Yes	Yes	FAP	Severe (>1000)	FAP	26	No	SWANOMA (39)	
025	HGBU	Patient	F	FAP	Yes	Brother CRC	AFAP	20	Mixed	18	No	Juvenile Gastric Polyps (45)	HYPERTHROIDISM (47)
026	HGBU	Patient	M	FAP	Yes	Yes	FAP	Severe (>1000)	FAP	20	CRC (20)	Duodenal Polyps	
028	HGBU	Patient	F	FAP	Yes	Uncle CRC / GrandMother Uterus / Mother Lung	FAP	Severe (>1000)	FAP	31	No	No	
036	HGBU	Patient	M	FAP	Yes	Mother CRC	FAP	Severe (>1000)	FAP	30	CRC (30)	No	
037	HGBU	Patient	F	FAP	Yes	Mother, Sister, Brother: CRC	FAP	Severe (>1000)	FAP	18	No	No	
038	HGBU	Patient	APC	F	FAP	Uncle Father Side ?	FAP	Severe (>1000)	FAP	25	No	Gastric Polyps	
039	HGBU	Patient	APC	F	FAP	Brother, Mother, Aunt: CRC	FAP	Severe (>1000)	FAP	10	CRC (46)	Gastric Duodenal Polyps (20)	
041	HGBU	Patient	M	FAP	No	Father, GrandFather: CRC	FAP	Severe (>1000)	FAP	25	CRC (25)	No	
044	HGBU	Patient	F	FAP	No	Father CRC	FAP	Severe (>1000)	Adenomas	45	CRC (45)	Gastric Duodenal Polyps (45)	
051	HGBU	Patient	F	FAP	Yes	Father, ?	FAP	Severe (>1000)	Adenomas	16	No	Gastric Polyps (16)	
054	HGBU	Patient	M	FAP	Yes	Brother CRC	FAP	Severe (>1000)	Adenomas	28	No	Gastric Polyps (28)	
056	HGBU	Patient	M	FAP	Yes	No	FAP	Severe (>1000)	Adenomas	40	No	No	
059	HGBU	Patient	APC	F	FAP	Mother CRC?	FAP	Severe (>1000)	Adenomas	22	No	No	
061	HGBU	Patient	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	24	No	No	
062	HGBU	Patient	M	FAP	No	Mother CRC	FAP	Severe (>1000)	Adenomas	48	CRC (48)	No	
063	HGBU	Patient	M	FAP	Yes	No	FAP	Severe (>1000)	Adenomas	32	No	Desmoid Tumor	Polyps: Gastric and Duodenum
064	HGBU	Patient	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	62	CRC (62)	Gastric Polyps	
065	HGBU	Patient	F	FAP	No	No	AFAP	Severe (>1000)	Adenomas	78	CRC (78)	No	
066	HGBU	Patient	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	15	No	Desmoid Tumor (26)	Polyps Duodenum (22)
067	HGBU	Patient	APC	M	FAP	Yes	FAP	Severe (>1000)	Adenomas	38	CRC (38)	Ostoma (35)	Gastric Polyps (38)
068	HGBU	Patient	M	FAP	Yes	CRC	FAP	Severe (>1000)	Adenomas	21	No	Ostoma (8)	
070	HGBU	Patient	F	FAP	No	Aunt - Grandfather CRC	FAP	Severe (>1000)	Adenomas	36	No	Desmoid Tumor (36)	Polyps Duodenum (36)
072	HGBU	Patient	NA	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	27	No	No	
073	HGBU	Patient	NA	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	No	No	No	
074	HGBU	Patient	NA	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	No	No	No	
076	HGBU	Patient	NA	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	No	No	No	
116	HIBA	Patient	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	59	No	No	
117	HIBA	Patient	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	54	No	No	
172	HEC	Patient	NA	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	No	No	No	
175	HEC	Patient	NA	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	No	No	No	
176	HEC	Patient	NA	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	No	No	No	
177	HEC	Patient	NA	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	No	No	No	
178	HEC	Patient	NA	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	No	No	No	
146	HEC	Patient	NA	FAP	Yes	Yes	NA	FAP	Adenomas	No	No	No	
151	HEC	Patient	NA	FAP	Yes	Yes	NA	FAP	Adenomas	No	No	No	
152	HEC	Patient	NA	FAP	Yes	Yes	NA	FAP	Adenomas	No	No	No	
153	HEC	Patient	NA	FAP	Yes	Yes	NA	FAP	Adenomas	No	No	No	
127	HIBA	Patient	M	FAP	No	No	AFAP	<100	Adenomas	64	No	No	
128	HIBA	Patient	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	18	No	No	
129	HIBA	Patient	F	FAP	No	Yes	FAP	Severe (>1000)	Adenomas	25	No	Gastric Polyps	
130	HIBA	Patient	M	FAP	No	No	AFAP	20	Adenomas	56	No	No	
131	HIBA	Patient	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	17	No	Gastric Polyps	
132	HIBA	Patient	M	FAP	No	No	AFAP	30	Adenomas	52	No	No	
133	HIBA	Patient	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	15	No	No	
134	HIBA	Patient	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	33	No	No	
135	HIBA	Patient	M	FAP	Yes	No	AFAP/Mixed	<100	Hiperplastic Adenomas	33	No	No	
136	HIBA	Patient	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	45	CRC (47)	Gastric Polyps (45)	
137	HIBA	Patient	M	FAP	No	Yes	FAP	Severe (>1000)	Adenomas	48	Rectum (48)	No	
138	HIBA	Patient	M	FAP	Yes	No	FAP	Severe (>1000)	Adenomas	32	No	No	
140	HIBA	Patient	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	62	CRC (62)	No	
141	HIBA	Patient	M	FAP	No	No	AFAP	<100	Adenomas	78	CRC (78)	No	
142	HIBA	Patient	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	15	No	No	
143	HIBA	Patient	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	38	CRC (38)	No	
144	HIBA	Patient	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	38	CRC (38)	No	
145	HIBA	Patient	F	FAP	Yes	Yes	AFAP	>100	Adenomas	24	No	No	
118	HIBA	Patient	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	50	CRC (50)	No	
119	HIBA	Patient	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	57	CRC (57)	No	
120	HIBA	Patient	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	54	CRC (54)	No	
121	HIBA	Patient	M	FAP	Yes	Yes	AFAP	<100	Adenomas	72	No	No	
122	HIBA	Patient	M	FAP	Yes	Yes	FAP	>100	Adenomas	62	CRC (62)	No	
123	HIBA	Patient	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	50	CRC (50)	No	
124	HIBA	Patient	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	31	CRC (31)	No	
125	HIBA	Patient	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	42	CRC (42)	No	
189	HIBA	Patient	M	FAP	No	Yes	AFAP	<50	Adenomas	58	CRC (58)	Thyroid Cancer (19)	
190	HIBA	Patient	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	26	No	No	
191	HIBA	Patient	F	FAP	No	No	FAP	Severe (>1000)	Adenomas	55	Yes (55)	Rectum (55)	Thyroid Nodule
192	HIBA	Patient	M	FAP	Yes	No	AFAP	<100	Adenomas	51	CRC (51)	Osteoma	Thyroid Nodule
193	HIBA	Patient	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	10	No	Duodenal Adenoma	
194	HIBA	Patient	F	FAP	No	No	FAP	Severe (>1000)	Adenomas	27	No	No	
195	HIBA	Patient	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	26	No	Swanoma (39)	
196	HIBA	Patient	F	FAP	Yes	Yes	AFAP	<50	Mixed	18	No	Gastric Polyps	Hyperthyroidism
197	HIBA	Patient	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	20	CRC (20)	Duodenal Polyps	
198	HIBA	Patient	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	31	No	No	
199	HIBA	Patient	M	FAP	No	No	AFAP	<100	Adenomas	64	No	No	
200	HIBA	Patient	M	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	18	No	No	
201	HIBA	Patient	F	FAP	No	Yes	FAP	Severe (>1000)	Adenomas	25	No	Gastric Polyps	
202	HIBA	Patient	M	FAP	No	Yes	AFAP	<100	Adenomas	56	No	No	
203	HGBU	Patient	F	FAP	Yes	Yes	FAP	Severe (>1000)	Adenomas	17	No	Gastric Polyps	

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Supplementary Table 52

Case		Gene	Gemline Identified Variant										Variant is NOVEL		ClinVar		
Case ID	Classification	Affected	Transcript	Chr	Position	HGVSc.	HGVSp.	Ref Allele	Alt Allele	Variant Type	Coding Impact	Zygosity	(Not Reported in ClinVar)	Variation ID	Preferred Name	Report Count	Significance
005	Index Patient	APC	NM_000038.6	5	11283962	c.4098_4111del	p.Gln327Serfs*3	TGACACACAAAA	A	Deletion	frameshift	Het	NOVEL (True)				
010	Index Patient	APC	NM_000038.6	5	11281904	c.1271dup	p.Glu245Glyfs*4	G	A	Insertion	frameshift	Het	NOVEL (True)				
014	Index Patient	APC	NM_000038.6	5	112838043	c.2450del	p.Gly817Alafs*3	G	T	Deletion	frameshift	Het	NOVEL (True)				
033	Index Patient	APC	NM_000038.6	5	11283953	c.4356del	p.Asn445Ilefs*18	T	T	Deletion	frameshift	Het	NOVEL (True)				
077	Relative to case 033	APC	NM_000038.6	5	11283953	c.4356del	p.Asn445Ilefs*18	T	T	Deletion	frameshift	Het	NOVEL (True)				
075	Index Patient	APC	NM_000038.6	5	11283546	c.2952dup	Ser583Ilefs*4	A	A	Insertion	frameshift	Het	NOVEL (True)				
088	Index Patient	APC	NM_000038.6	5	11283814	c.2520dup	p.Ser1395Lysfs*3	C	A	Insertion	frameshift	Het	NOVEL (True)				
023	Index Patient	APC	NM_000038.6	5	11283978	c.4183dup	p.Ser1395Lysfs*3	A	A	Insertion	frameshift	Het	NOVEL (True)				
174	Index Patient	APC	NM_000038.6	5	11283978	c.4183dup	p.Ser1395Lysfs*3	A	A	Insertion	frameshift	Het	NOVEL (True)				
185	Relative to case 170	APC	NM_000038.6	5	11283978	c.4183dup	p.Ser1395Lysfs*3	C	A	Insertion	frameshift	Het	NOVEL (True)				
186	Relative to case 170	APC	NM_000038.6	5	11283978	c.4183dup	p.Ser1395Lysfs*3	C	A	Insertion	frameshift	Het	NOVEL (True)				
187	Relative to case 174	APC	NM_000038.6	5	11283978	c.4183dup	p.Ser1395Lysfs*3	C	A	Insertion	frameshift	Het	NOVEL (True)				
188	Relative to case 174	APC	NM_000038.6	5	11283978	c.4183dup	p.Ser1395Lysfs*3	C	A	Insertion	frameshift	Het	NOVEL (True)				
019	Index Patient	ERCC4	NM_005236.6	16	13939090	c.1795G>A	p.A298G	G	A	SNV	missense	Het	True				
046	Index Patient	NELL1	NM_001256552.1	15	75349153	c.506G>A	p.Gly169Asp	G	A	SNV	missense	Het	True				
002	Index Patient	APC	NM_000038.6	5	11283937	c.3803dup	p.Ile1269Aunfs*7	C	A	Insertion	frameshift	Het	False	2682626	NM_000038.6(APC):c.3803dup (p.Ile1269A)	2	Pathogenic
011	Index Patient	APC	NM_000038.6	5	11283991	c.3758dup	p.Glu1257Argfs*19	C	A	Insertion	frameshift	Het	False	2126315	NM_000038.6(APC):c.3758dup (p.Glu1257A)	2	Pathogenic
003	Index Patient	APC	NM_000038.6	5	112837913	c.2319del	p.Phe773Leufs*4	T	A	Deletion	frameshift	Het	False	1073520	NM_000038.6(APC):c.2319del (p.Phe773L)	2	Pathogenic
016	Index Patient	APC	NM_000038.6	5	11283839	c.2805C>A	p.Tyr935*	C	A	SNV	nonsense	Het	False	810	NM_000038.6(APC):c.2805C>A (p.Tyr935T)	20	Pathogenic
020	Index Patient	APC	NM_000038.6	5	11283894	c.2802_2805del	p.Tyr935Ilefs*19	ACTT	TT	Deletion	frameshift	Het	False	42956	NM_000038.6(APC):c.2802_2805del (p.Tyr935I)	6	Pathogenic
030	Index Patient	APC	NM_000038.6	5	11283915	c.3927_3931del	p.Glu1309Aspfs*4	AAAGA	GA	Deletion	frameshift	Het	False	816	NM_000038.6(APC):c.3927_3931del (p.Glu1309A)	44	Pathogenic
031	Index Patient	APC	NM_000038.6	5	11283915	c.3927_3931del	p.Glu1309Aspfs*4	AAAGA	GA	Deletion	frameshift	Het	False	1482312	NM_000038.6(APC):c.2016_2017del (p.H6672fs)	4	Pathogenic
032	Index Patient	APC	NM_000038.6	5	11280312	c.763dup	p.H625Pprofs*2	AT	AG	Insertion	frameshift	Het	False	3213188	NM_000038.6(APC):c.763dup (p.H625Pfs)	1	Pathogenic
034	Index Patient	APC	NM_000038.6	5	11284026	c.612_401del	p.C412_401del	C	A	Deletion	frameshift	Het	False	823	NM_000038.6(APC):c.465_463del (p.Glu1538fs)	10	Pathogenic
035	Index Patient	APC	NM_000038.6	5	11275711	c.509_512del	p.Asp170Valfs*4	ATAG	ATAG	Deletion	frameshift	Het	False	804	NM_000038.6(APC):c.509_512del (p.Asp170fs)	14	Pathogenic
038	Index Patient	APC	NM_000038.6	5	11283915	c.3927_3931del	p.Glu1309Aspfs*4	AAAGA	GA	Deletion	frameshift	Het	False	816	NM_000038.6(APC):c.3927_3931del (p.Glu1309A)	44	Pathogenic
039	Index Patient	APC	NM_000038.6	5	11283915	c.3927_3931del	p.Glu1309Aspfs*4	AAAGA	GA	Deletion	frameshift	Het	False	816	NM_000038.6(APC):c.3927_3931del (p.Glu1309A)	44	Pathogenic
040	Index Patient	APC	NM_000038.6	5	11283915	c.3927_3931del	p.Glu1309Aspfs*4	AAAGA	GA	Deletion	frameshift	Het	False	127281	NM_000038.6(APC):c.3412C>T (p.Arg805Ter)	14	Pathogenic
042	Index Patient	APC	NM_000038.6	5	11283914	c.3920T>A	p.Ile1307Lys	T	A	SNV	missense	Het	False	822	NM_000038.6(APC):c.3920T>A (p.Ile1307Lys)	53	Conflicting classifications of pathogenicity; association; risk factor
045	Index Patient	APC	NM_000038.6	5	11283839	c.2795C>G	p.Ser932*	C	G	SNV	nonsense	Het	False	236581	NM_000038.6(APC):c.2795C>G (p.Ser932Ter)	2	Pathogenic
047	Index Patient	APC	NM_000038.6	5	11283874	c.3183_3187del	p.Gln1062_Ser1063delins*	AAAAC	AAAC	Deletion	nonsense	Het	False	88913	NM_000038.6(APC):c.3183_3187del (p.Gln1062_Ser1063delins)	31	Pathogenic
050	Index Patient	APC	NM_000038.6	5	11283977	c.3202C>G	p.Ser968*	C	G	SNV	missense	Het	False	490258	NM_000038.6(APC):c.3202C>G (p.Ser968Ter)	2	Pathogenic
052	Index Patient	APC	NM_000038.6	5	11283911	c.3920_3924del	p.Ile1307Argfs*6	AAATA	T	SNV	frameshift	Het	False	419999	NM_000038.6(APC):c.3920_3924del (p.Ile1307fs)	2	Pathogenic
055	Index Patient	APC	NM_000038.6	5	112840023	c.4429C>T	p.C4429C>T	C	T	SNV	nonsense	Het	False	2584019	NM_000038.6(APC):c.4429C>T (p.C4429T)	1	Pathogenic
057	Index Patient	APC	NM_000038.6	5	11283937	c.3783_3784del	p.Tyr1261Leufs*13	TT	TT	Deletion	frameshift	Het	False	537503	NM_000038.6(APC):c.3783_3784del (p.Tyr1261fs)	10	Pathogenic
058	Index Patient	APC	NM_000038.6	5	11283789	c.2396_2397del	p.Tyr799Cysfs*3	TA	TA	Deletion	frameshift	Het	False	2502365	NM_000038.6(APC):c.2396_2397del (p.Tyr799fs)	3	Pathogenic/Likely pathogenic
059	Index Patient	APC	NM_000038.6	5	11283915	c.3927_3931del	p.Glu1309Aspfs*4	AAAGA	GA	Deletion	frameshift	Het	False	816	NM_000038.6(APC):c.3927_3931del (p.Glu1309A)	44	Pathogenic
067	Index Patient	APC	NM_000038.6	5	11283915	c.3927_3931del	p.Glu1309Aspfs*4	AAAGA	GA	Deletion	frameshift	Het	False	816	NM_000038.6(APC):c.3927_3931del (p.Glu1309A)	44	Pathogenic
069	Index Patient	APC	NM_000038.6	5	11283915	c.3927_3931del	p.Glu1309Aspfs*4	AAAGA	GA	Deletion	frameshift	Het	False	816	NM_000038.6(APC):c.3927_3931del (p.Glu1309A)	44	Pathogenic
071	Index Patient	APC	NM_000038.6	5	11283829	c.3336_3340del	p.Asn113Serfs*4	CAAT	A	Deletion	frameshift	Het	False	826233	NM_000038.6(APC):c.3336_3340del (p.Asn113fs)	3	Pathogenic
079	Index Patient	APC	NM_000038.6	5	11283877	c.3193C>A	p.Gln1065Lys	C	A	SNV	nonsense	Het	False	469914	NM_000038.6(APC):c.3193C>A (p.Gln1065Lys)	6	Uncertain significance
080	Index Patient	APC	NM_000038.6	5	11283859	c.2959C>G	p.Gln1069Ser	G	G	SNV	nonsense	Het	False	584668	NM_000038.6(APC):c.2959C>G (p.Gln1069Ser)	4	Uncertain significance
081	Index Patient	APC	NM_000038.6	5	11283837	c.2805A>G	p.Tyr935*	C	G	SNV	nonsense	Het	False	93344	NM_000038.6(APC):c.2805A>G (p.Tyr935Ter)	4	Pathogenic
082	Index Patient	APC	NM_000038.6	5	11283914	c.3920T>A	p.Ile1307Lys	T	A	SNV	missense	Het	False	822	NM_000038.6(APC):c.3920T>A (p.Ile1307Lys)	53	Conflicting classifications of pathogenicity; association; risk factor
083	Index Patient	APC	NM_000038.6	5	11284375	c.814G>A	p.Arg274His	G	A	SNV	missense	Het	False	414178	NM_000038.6(APC):c.814G>A (p.Arg274His)	11	Conflicting classifications of pathogenicity
084	Index Patient	APC	NM_000038.6	5	11283949	c.174A_2A>G	p.S174A_2A>G	A	G	Splicing Deletion	frameshift	Het	False	155482	NM_000038.6(APC):c.174A_2A>G (p.S174A_2A>G)	9	Pathogenic
085	Index Patient	APC	NM_000038.6	5	11283815	c.2522del	p.Leu841*	L	SNV	nonsense	Het	False	642670	NM_000038.6(APC):c.2522del (p.Leu841fs)	1	Pathogenic	
086	Index Patient	APC	NM_000038.6	5	11283792	c.2333G>A	p.Asn778Ser	A	G	SNV	nonsense	Het	False	630116	NM_000038.6(APC):c.2333G>A (p.Asn778Ser)	3	Uncertain significance
087	Index Patient	APC	NM_000038.6	5	11284010	c.4508C>A	p.Ser1503*	C	A	SNV	nonsense	Het	False	2584283	NM_000038.6(APC):c.4508C>A (p.Ser1503Ter)	1	Pathogenic
089	Index Patient	APC	NM_000038.6	5	11283839	c.2805C>A	p.Tyr935*	C	A	SNV	nonsense	Het	False	810	NM_000038.6(APC):c.2805C>A (p.Tyr935Ter)	20	Pathogenic
090	Index Patient	APC	NM_000038.6	5	11283955	c.4361A>T	p.Lys1454Ile	A	T	SNV	nonsense	Het	False	630564	NM_000038.6(APC):c.4361A>T (p.Lys1454Ile)	2	Uncertain significance
091	Index Patient	APC	NM_000038.6	5	11283874	c.3183_3187del	p.Gln1062_Ser1063delins*	AAAAC	AAAC	Deletion	nonsense	Het	False	88913	NM_000038.6(APC):c.3183_3187del (p.Gln1062_Ser1063delins)	31	Pathogenic
092	Index Patient	APC	NM_000038.6	5	11283922	c.4128T>A	p.Tyr1376*	T	A	SNV	nonsense	Het	False	178059	NM_000038.6(APC):c.4128T>A (p.Tyr1376Ter)	1	Pathogenic
093	Index Patient	APC	NM_000038.6	5	11283915	c.3927_3931del	p.Glu1309Aspfs*4	AAAG	GA	Deletion	frameshift	Het	False	816	NM_000038.6(APC):c.3927_3931del (p.Glu1309A)	44	Pathogenic
094	Index Patient	APC	NM_000038.6	5	11283926	c.3940C>T	p.Arg324*	C	T	SNV	nonsense	Het	False	184937	NM_000038.6(APC):c.3940C>T (p.Arg324Ter)	15	Pathogenic
095	Index Patient	APC	NM_000038.6	5	11283900	c.1619_1620del	p.Gln14Aspfs*9	GG	T	Insertion	frameshift	Het	False	2584191	NM_000038.6(APC):c.1619_1620del (p.Gln14fs)	1	Pathogenic
096	Index Patient	APC	NM_000038.6	5	11283967	c.4025dup	p.Leu1342Phefs*12	GG	T	Insertion	frameshift	Het	False	217976	NM_000038.6(APC):c.4025dup (p.Leu1342fs)	6	Pathogenic
097	Index Patient	APC	NM_000038.6	5	11283940	c.3815C>G	p.Ser1272*	C	G	SNV	nonsense	Het	False	217973	NM_000038.6(APC):c.3815C>G (p.Ser1272Ter)	6	Pathogenic
098	Index Patient	APC	NM_000038.6	5	11283992	c.4961dup	p.Met321Aunfs*6	C	A	Insertion	frameshift	Het	False	2774690	NM_000038.6(APC):c.4961dup (p.Met321fs)	1	Pathogenic
100	Index Patient	APC	NM_000038.6	5	11283914	c.3920T>A	p.Ile1307Lys	T	A	SNV	missense	Het	False	822	NM_000038.6(APC):c.3920T>A (p.Ile1307Lys)	53	Conflicting classifications of pathogenicity; association; risk factor
101	Index Patient	APC	NM_000038.6	5	11284205	c.6431T>C	p.Leu2144Pro	T	C	SNV	nonsense	Het	False	187631	NM_000038.6(APC):c.6431T>C (p.Leu2144Pro)	5	Uncertain significance
102	Index Patient	APC	NM_000038.6	5	11283922</												

SUPPLEMENTARY TABLE S3

studyld	novel_pos	n_hits
msk_chord_2024	1367	101
msk_chord_2024	1395	94
msk_met_2021	1367	72
msk_met_2021	1395	63
pancan_pdmr_2025	1367	41
pancan_mimsi_msk_2024	1367	37
pancan_mimsi_msk_2024	1395	36
pancan_pdmr_2025	1395	33
msk_chord_2024	1455	32
crc_eo_2020	1367	31
rectal_msk_2022	1395	26
msk_met_2021	1455	24
crc_eo_2020	1395	23
crc_msk_2017	1367	18
crc_msk_2017	1395	18
pancan_mimsi_msk_2024	1455	17
rectal_msk_2022	1367	14
coadread_dfc_i_2016	1395	13
coadread_tcga_pan_can_atlas_2018	1395	12
rectal_msk_2019	1395	12
crc_eo_2020	1455	11
crc_apc_impact_2020	1395	10
coadread_dfc_i_2016	1367	9
msk_chord_2024	425	9
coadread_tcga_pan_can_atlas_2018	1367	8
msk_chord_2024	985	8
msk_met_2021	985	7
coad_tcga_gdc	1367	6
coad_tcga_gdc	1395	6
crc_apc_impact_2020	1367	6
crc_msk_2017	1455	6
pancan_pdmr_2025	1455	6
coad_silu_2022	1395	5
pancan_mimsi_msk_2024	985	5
rectal_msk_2019	1367	5
coad_tcga_gdc	1455	4
coadread_tcga	1367	4
coadread_tcga_pub	1367	4
crc_apc_impact_2020	1455	4
msk_chord_2024	842	4
msk_met_2021	425	4

pancan_mimsi_msk_2024	425	4
prad_pik3r1_msk_2021	1455	4
read_tcga_gdc	1367	4
read_tcga_gdc	1395	4
rectal_msk_2022	985	4
coad_silu_2022	1367	3
coadread_dfcf_2016	1455	3
coadread_tcga_pan_can_atlas_2018	1455	3
prad_msk_stopsack_2021	1455	3
prostate_msk_2024	1455	3
tmb_mskcc_2018	1395	3
coad_cptac_2019	1367	2
coad_cptac_gdc	1367	2
coad_tcga_gdc	985	2
coadread_cass_2020	1367	2
coadread_mskcc	1367	2
coadread_mskcc	1395	2
coadread_tcga_pan_can_atlas_2018	425	2
coadread_tcga_pan_can_atlas_2018	985	2
crc_apc_impact_2020	985	2
crc_eo_2020	985	2
crc_msk_2017	425	2
msk_access_2021	1455	2
msk_ctdna_vte_2024	1455	2
msk_met_2021	842	2
nslc_ctdx_msk_2022	1455	2
pancan_mimsi_msk_2024	842	2
pancan_pcawg_2020	1395	2
prad_cdk12_mskcc_2020	1455	2
rectal_msk_2019	1455	2
rectal_msk_2022	425	2
stad_tcga_pub	1455	2
tmb_mskcc_2018	1367	2
blca_msk_tcga_2020	985	1
blca_tcga_gdc	985	1
blca_tcga_pan_can_atlas_2018	985	1
blca_tcga_pub_2017	985	1
brca_tcga	425	1
brca_tcga_pub2015	425	1
coad_caseccc_2015	1395	1
coad_cptac_2019	842	1
coad_cptac_2019	1455	1
coad_cptac_gdc	842	1
coad_cptac_gdc	1455	1

coad_silu_2022	425	1
coad_silu_2022	1455	1
coad_tcga_gdc	425	1
coadread_cass_2020	985	1
coadread_cass_2020	1395	1
coadread_dfc_2016	425	1
coadread_dfc_2016	842	1
coadread_genentech	1367	1
coadread_tcga	985	1
coadread_tcga	1395	1
coadread_tcga	1455	1
coadread_tcga_pub	985	1
coadread_tcga_pub	1395	1
coadread_tcga_pub	1455	1
crc_dd_2022	1455	1
crc_eo_2020	425	1
crc_eo_2020	842	1
crc_hta11_htan_2021	985	1
crc_msk_2017	985	1
crc_nigerian_2020	1455	1
crc_orion_2024	1395	1
egc_msk_2023	1455	1
kich_tcga	425	1
msk_chord_2024	817	1
msk_ctdna_vte_2024	1367	1
pancan_pcawg_2020	985	1
prad_mcspc_mskcc_2020	1455	1
prad_pik3r1_msk_2021	817	1
prad_pik3r1_msk_2021	842	1
prostate_msk_2024	817	1
prostate_msk_2024	842	1
read_tcga_gdc	425	1
read_tcga_gdc	1455	1
rectal_msk_2019	985	1
rectal_msk_2022	1455	1
rectal_radiation_msk_2024	1367	1
stad_tcga	1455	1
stad_tcga_gdc	1455	1
stad_tcga_pan_can_atlas_2018	1455	1
stes_tcga_pub	1455	1
tmb_mskcc_2018	425	1
tmb_mskcc_2018	1455	1

Supplementary Table S4

Oligo name	Region (1)	Forward (5'→3')	Reverse (5'→3')	Fragment size (bp)
APCex2	Exon 2	TTTCTTTAAAAACAAGCAGCCA	CACAGAAAACCTTGCCTCAG	519
APCex3	Exon 3	AAGGTGCGTGCTTTGAGAGT	ACCAACACCCAAATCGAGAG	308
APCex4	Exon 4	CCAAGTGGACTTTTCAGGGA	CTGGAGTACACAAGGCAATGTT	412
APCex5	Exon 5	GCTCTTCTGCAGTCTTTATTAGCA	CCTAGTTGAACCCTGAGGTCC	444
APCex6	Exon 6	AAGCCACTTGTGACTTTGGC	GTTGCTCAGCAGCCATGATA	511
APCex7	Exon 7	TGCGGTGAGCTGAGATTATG	ACCCACAAACAAGAAAGGCA	376
APCex8	Exon 8	GCAGCTCTAATGCTCAAGGG	TGGTACTGAATGCTTCTGGAAA	465
APCex9	Exon 9	CCATTCTGCAGTTTAATGCTCA	TAGAGATGGGGTTTTGCCAC	512
APCex10	Exon 10	CTGGAAGGTTTTCCGGTTT	TGCTTTGAAACATGCACTACG	577
APCex11	Exon 11	GTCAAGGGCAGATGAGTGGT	TTCTATGCTGGAACCAGGG	511
APCex12	Exon 12	TTGTCTTTTTCTCTTGCCC	AGCGAATGTGAAGCACAGGT	362
APCex13	Exon 13	CCTGTTGCTTATCATTCTCACC	AGAGTGAGACCCTGCCTCAA	391
APCex14	Exon 14	CAGCCTCCCAAAGTGATAGG	ATGGCTAAAAGAAGGCAGCA	341
APCex15	Exon 15	AGGGACGGGCAATAGGATAG	CATTGCTTACAATTAGGTCTTTTGA	404
APCex15_12	Exon 16	TACTGCATACACATTGTGACCTT	CGAGGGTTTCATTTGACCTC	1040
APCex15_34	Exon 16	CAATTTCACTAAGTCGGAAAATTCA	CTGCTTCCTGTGTGCTGCTGA	1085
APCex15_516	Exon 16	GCTCCATCCAAGTTCTGCAC	CATGGTTTGTCCAGGGCTAT	604
APCex15_156	Exon 16	GGCATTATAAGCCCCAGTGA	ACTTGGTTTCTTGCCACAG	585

(1) According to ensemble transcript ENST00000257430.9