

Supplementary Materials: Results and Clinical Interpretation of Germline RET Analysis in A Series of Patients with Medullary Thyroid Carcinoma: The Challenge of the Variants of Uncertain Significance.

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Table S1. Case series under study.

Patient number	Family ID	Family phenotype	Affected/Healthy	Type of analysis	RET variant
1	10-O-02	MTC	A	C	NO
2	28-O-03	MTC	A	C	NO
3	91-O-03	MTC	A	C	YES
4	91-O-03	MTC	A	T	YES
5	31-O-04	MTC	A	C	NO
6	144-O-04	MTC	A	C	YES
7	59-M-04	MTC	A	C	NO
8	91-O-03	MTC	A	T	YES
9	91-O-03	MTC	A	T	YES
10	91-O-03	MTC	H	T	YES
11	91-O-03	MTC	H	T	NO
12	91-O-03	MTC	H	T	NO
13	27-O-05	MTC	A	C	NO
14	91-O-03	MTC	H	T	YES
15	91-O-03	MTC	A	T	YES
16	91-O-03	MTC	H	T	NO
17	42-O-07	MTC	A	C	NO
18	9-O-08	MTC	H	T	YES
19	LAB-01	UK	UK	C	NO
20	LAB-02	UK	UK	C	NO
21	LAB-03	IG	UK	C	NO
22	9-O-08	MTC	H	T	NO
23	9-O-08	MTC	H	T	YES
24	91-O-03	MTC	H	T	NO
25	19-O-09	MTC	A	C	NO
26	LAB-04	UK	A	C	NO
27	21-O-09	MTC	A	C	NO
28	31-O-09	MTC	A	C	NO
29	54-O-09	MTC	A	C	YES
30	54-O-09	MTC	A	T	YES
31	42-O-10	MTC	A	C	NO

32	91-O-03	MTC	H	T	NO
33	13-O-10	MTC	A	C	NO
34	44-O-10	MTC	A	C	NO
35	53-O-10	MTC	A	C	NO
36	63-O-10	MTC	A	C	NO
37	54-O-09	MTC	H	T	YES
38	54-O-09	MTC	H	T	YES
39	12-O-11	MTC	A	C	NO
40	13-O-11	MEN2A	A	C	YES
41	29-O-11	MTC	A	C	NO
42	33-O-11	MTC	A	C	NO
43	53-O-11	MTC	A	C	NO
44	67-O-11	MTC	A	C	NO
45	68-O-11	MTC	A	C	YES
46	92-O-11	MTC	A	C	NO
47	74-O-11	CCH	A	C	NO
48	87-O-11	MTC	A	C	NO
49	13-O-12	MTC	A	C	NO
50	54-O-11	MTC	A	C	NO
51	35-O-12	CCH	A	C	NO
52	190-O-13	PHPT	A	C	NO
53	190-O-13	PHPT	A	C	NO
54	58-O-12	MTC	A	C	NO
55	65-O-12	MTC	A	C	NO
56	100-O-12	MTC	A	C	NO
57	LAB-05	PHPT	UK	C	NO
58	228-O-18	MTC	A	C	YES
59	LAB-06	PGL/PCC	A	C	NO
60	58-O-13	MTC	A	C	NO
61	53-O-13	PHPT	A	C	NO
62	72-O-13	MTC	H	T	YES
63	136-O-13	PHPT	A	C	YES
64	81-O-13	MTC	A	C	NO
65	89-O-13	MTC	A	C	NO
66	54-O-09	MTC	H	T	YES
67	103-O-13	MTC	A	C	NO
68	LAB-07	PHPT	UK	C	NO
69	121-O-13	MTC	A	C	NO
70	228-O-18	MTC	A	T	YES
71	228-O-18	MTC	A	T	YES
72	228-O-18	MTC	H	T	YES
73	228-O-18	MTC	A	T	YES
74	228-O-18	MTC	H	T	NO

75	10-O-14	MTC	A	C	NO
76	81-O-14	MTC	A	C	NO
77	228-O-18	MTC	H	T	YES
78	228-O-18	MTC	H	T	NO
79	228-O-18	MTC	H	T	NO
80	144-O-14	MTC	A	C	NO
81	168-O-14	MTC	A	C	YES
82	175-O-14	MTC	A	C	NO
83	LAB-08	PTC	UK	C	NO
84	204-O-14	MTC	A	C	NO
85	168-O-14	MTC	H	T	YES
86	120-O-14	PGL/PCC	A	C	NO
87	LAB-09	UK	UK	C	NO
88	LAB-10	UK	UK	C	NO
89	269-O-14	MTC	A	C	NO
90	LAB-11	CCH	UK	C	NO
91	168-O-14	MTC	H	T	YES
92	168-O-14	MTC	H	T	YES
93	168-O-14	MTC	H	T	NO
94	LAB-12	UK	UK	T	YES
95	LAB-13	PHPT	UK	C	NO
96	31-O-15	PGL/PCC	A	C	NO
97	39-O-15	MTC	A	C	NO
98	45-O-15	PGL/PCC	A	C	NO
99	53-O-15	PGL/PCC	A	C	NO
100	89-O-15	MTC	A	C	YES
101	89-O-15	MTC	H	T	NO
102	89-O-15	MTC	H	T	YES
103	89-O-15	MTC	H	T	YES
104	89-O-15	MTC	H	T	YES
105	169-O-15	MEN2B	A	C	YES
106	169-O-15	MEN2B	H	T	NO
107	169-O-15	MEN2B	H	T	NO
108	169-O-15	MEN2B	H	T	NO
109	169-O-15	MEN2B	H	T	NO
110	169-O-15	MEN2B	H	T	NO
111	32-O-15	MTC	A	C	NO
112	70-O-15	PGL/PCC	A	C	NO
113	219-O-15	PGL/PCC	A	C	NO
114	316-O-15	PGL/PCC	A	C	NO
115	13-O-16	MTC	A	C	NO
116	121-O-16	PGL/PCC	A	C	NO
117	LAB-14	UK	UK	C	NO

118	LAB-15	UK	UK	C	NO
119	LAB-16	UK	UK	C	YES
120	LAB-17	UK	UK	C	NO
121	204-O-16	PGL/PCC	A	C	NO
122	275-O-16	MTC	A	C	NO
123	371-O-16	CCH	A	C	NO
124	166-O-17	MTC	A	C	NO
125	191-O-17	MTC	H	T	YES
126	288-O-17	PGL/PCC	A	C	NO
127	359-O-17	PGL/PCC	A	C	NO
128	1-MAG-CRC	PGL/PCC	A	C	NO
129	LAB-18	UK	UK	C	NO
130	43-O-18	MTC	A	C	NO
131	55-O-18	MTC	A	C	YES
132	55-O-18	MTC	H	T	NO
133	LAB-19	UK	UK	C	NO
134	LAB-20	MEN2B	A	C	YES
135	LAB-20	MEN2B	H	T	NO
136	LAB-20	MEN2B	H	T	NO
137	55-O-18	MTC	H	T	NO
138	55-O-18	MTC	H	T	NO
139	LAB-21	UK	UK	C	YES
140	LAB-21	UK	UK	T	NO
141	LAB-21	UK	UK	T	NO
142	142-O-18	MEN2A	A	T	YES
143	142-O-18	MEN2A	H	T	NO
144	142-O-18	MEN2A	H	T	NO
145	LAB-22	UK	UK	C	NO
146	228-O-18	MTC	A	T	YES
147	228-O-18	MTC	H	T	YES
148	228-O-18	MTC	H	T	NO
149	228-O-18	MTC	H	T	NO
150	228-O-18	MTC	A	T	YES
151	LAB-23	UK	UK	C	NO
152	LAB-24	UK	UK	C	NO
153	LAB-25	UK	UK	C	NO
154	LAB-26	UK	UK	C	NO
155	LAB-27	UK	UK	T	NO
156	LAB-28	UK	UK	C	NO
157	228-O-18	MTC	H	T	NO
158	520-M-18	MTC	A	C	NO
159	324-O-19	MTC	A	C	NO
160	330-O-19	PGL/PPC	A	C	NO

161	83-OB-19	PHPT	A	C	NO
162	115-O-20	MTC	H	T	YES
163	91-O-03	MTC	H	T	YES

Abbreviations: MTC = Medullary Thyroid Carcinoma; UK = Unknown; IG = Intestinal Ganglioneuromatosis; MEN2A = Multiple Endocrine Neoplasia type 2 A; CCH = C Cell Hyperplasia; PGL/PCC = Paraganglioma/Pheocromocytoma; PHPT = Primary Hyperparathyroidism; MEN2B = Multiple Endocrine Neoplasia type 2 B; C = Complete analysis; T = Targeted analysis