

Supplementary Data

Supplementary Table S1. Patient cohort, molecular genetics, and propionate incorporation

Patient	Age (years)	Sex	Subtype	Allele 1	Allele 2	Patient ¹⁴ C-propionate incorporation (nmol/mg protein/18 hr)		Control ¹⁴ C-propionate incorporation (nmol/mg protein/18 hr)	
						-OHcbl	+OHcbl	-OHcbl	+OHcbl
1	2.3	M	<i>mut^r</i>	c.1846C>T p.R616C	c.1846C>T p.R616C				
2	2.4	F	<i>mut^r</i>	c.91C>T p.R31X	c.323G>A p.R108H	2.10	3.10	15.50	20.00
3	2.5	M	<i>mut^r</i>	c.277C>T p.R93C	c.1106G>A p.R369H				
4	3.2	M	<i>mut^r</i>	c.1207C>T p.R403X	c.2150G>T p.G717V				
5	3.3	M	<i>mut^p</i>	c.1399C>T p.R467X	c.693C>G p.Y231X				
6	3.5	F	<i>mut^r</i>	c.281G>T p.G94V	c.2150G>T p.G717V				
7	4.2	F	<i>mut^p</i>	c.682C>T p.R228X	c.1106G>A p.R369H	0.40	0.40	19.40	24.00
8	4.4	M	<i>mut^p</i>	c.91C>T p.R31x	c.1181dupT p.L394FfsX30				
9	4.4, 2.3	F	<i>mut^p</i>	c.607G>A p.G203R	c.682C>T p.R228X	0.33	0.46	10.00	11.00
10	4.5	F	<i>mut^p</i>	c.1207C>T p.R403X	c.2008G>C p.G670R	1.10	1.30	12.50	13.50
11	4.6	F	<i>mut^p</i>	c.2080C>T p.R694V	c.643G>T p.G215C				
12	5.6	M	<i>mut^p</i>	c.322C>T p.R108C	c.682C>T p.R228X	1.40	1.50	15.80	20.10
13	6.0	F	<i>mut^p</i>	c.323G>A p.R108H	c.1867G>C p.G623R	0.28	0.28	3.10	7.30
14	6.1	M	<i>mut^p</i>	c.1741C>T p.R581x	c.753 + 2T>A p.?	0.26	0.33	11.40	12.60
15	6.5	F	<i>mut^p</i>	c.878A>C p.Q293P	c.878A>C p.Q293P				
16	6.6	M	<i>mut^p</i>	c.2179C>T p.R727X	c.2179C>T p.R727X				
17	7.2	M	<i>mut^p</i>	c.281G>T p.G94V	c.1867G>C p.G623R	1.30	1.50		
18	7.8	M	<i>mut^p</i>	c.1106G>A p.R369H	c.1778_1782delAAAGT p.S594RfsX11	1.85	2.52	24.40	26.00
19	8.6	M	<i>mut^p</i>	c.1022dupA p.N341KfsX17	c.671_678dup p.V227NfsX16	0.56	0.61		
20	9.4	M	<i>mut^p</i>	c.1105C>T p.R369C	c.1207C>T p.R403X	0.58	0.58		
21	9.7, 8.2	M	<i>mut^p</i>	c.1038_1040delTCT p.347delL	c.349G>T p.E117X				
22	10.4	F	<i>mut^p</i>	c.2179C>T p.R727X	c.2179C>T p.R727X	0.79	0.78	14.00	14.00
23	10.6	M	<i>mut^p</i>	c.670G>T p.E224X	c.682C>T p.R228X	0.80	0.80	10.30	12.50
24	10.9	M	<i>mut^p</i>	c.322C>T p.R108C	c.322C>T p.R108C	0.31	0.34	7.30	7.70
25	11.3	M	<i>mut^p</i>	c.682C>T p.R228X	c.682C>T p.R228X	0.14	0.13	6.00	6.00
26	11.8, 4.6	F	<i>mut^p</i>	c.1942G>C p.G642R	c.1942G>C p.G642R	1.60	1.80	15.00	15.00
27	11.9, 8.0	F	<i>mut^p</i>	c.927G>A p.W309X	c.983T>C p.L328P				
28	13.9, 11.9	M	<i>mut^p</i>	c.1106G>A p.R369H	c.1106G>A p.R369H	0.24	0.20	7.50	7.50
29	13.2, 8	F	<i>mut^p</i>	c.2053dupCTC p.685insL	c.91C>T p.R31X	0.96	1.30	12.00	16.00
30	17, 10.5, 12.6	F	<i>mut^p</i>	c.1658delT p.V553GfsX17	c.29dupT p.L111TfsX38	0.50	0.50	8.50	10.50
31	18.5, 16.4	M	<i>mut^p</i>	c.572C>A p.A191E	c.655A>T p.N219Y	2.10	1.70		
32	22.5	M	<i>mut^p</i>	c.322C>T p.R108C	c.1106G>A p.R369H	0.43	0.43	10.0	10.0
33	22.7, 24	M	<i>mut^p</i>	c.572C>A p.A191E	c.682C>T p.R228X	1.50	1.50	12.50	16.80
34	24.7	M	<i>mut^p</i>	c.655A>T p.N219Y	c.1048C>T p.H350Y				
35	26.8, 19.9	F	<i>mut^p</i>	c.1106G>A p.R369H	c.1106G>A p.R369H				
36	25.4, 25.4	F	<i>mut^p</i>	c.935G>T p.G312V	c.1909G>A p.G637R	1.30	1.20	15.00	15.70
37	26.1, 24.2	M	<i>mut^p</i>	c.1741C>T p.R581X	c.1741C>T p.R581X	1.40	1.10	13.50	14.60
38	26.5, 25.9	M	<i>mut^r</i>	c.1760A>C p.Y587S	c.2150G>T p.G717V				
39	27.5, 20.6	F	<i>mut^p</i>	c.2053dupCTC p.685insL	c.2053dupCTC p.685insL	0.80	0.90		
40	28.5	F	<i>mut^r</i>	c.2150G>T p.G717V	c.2150G>T p.G717V	0.70	1.90	20.90	20.80
41	28.7	F	<i>mut^r</i>	c.2150G>T p.G717V	unknown				
42	31.6, 27.5	F	<i>mut^p</i>	c.826G>T p.E276x	c.1106G>A p.R369H				