資料

ATR-X 症候群　日本人患者さんの 遺伝子変異のまとめ

Patient #	Family #	Exon/intron	Nucleotide change	Amino acid change	HbH
1	1	Ex1del	MAGT exon1-ATRX exon 1 large deletion		+
2	2	5'-UTR			+
3	3	2	Ex2_5		+
4			Ex2_5		+
5	4	INT2	(+)23A>G	?	
6	5	5	c.370G>T;r[243_484del]	p.R81fs	-
7		5	c.370G>T;r[243_484del]	p.R81fs	-
8	6	6	c.390_391 ins A	p.E131fs	
9	7			p.Pro170Leu	-
10	8	8	c.536A>G; r[532_594del]	p.V178_K198del	+
11	9	8	c.536A>G; r[532_594del]	p.V178_K198del	+
12	10	8	c.536A>G; r[532_594del]	p.V178_K198del	
13	11	8	c.536A>G; r[532_594del]	p.V178_K198del	+
14	12	8		p.P190L	+
15	13	8	c.569C>T	p.P190L	+
16	14	8	c.580G>A	p.V194I	+
17	15	8	c.581T>C	p.V194A	-
18	16	10	c.667T>C	p.C223R	-
19	17	10	c.668G>T	p.C223F	+
20	18	10	c.687G>C	p.L229F	+
21	19	10	c.695G>A	p.C232Y	
22	20	10	c.727T>C	p.C243R	
23	21	10	c.734T>C	p.L245P	-
24		10	c.734T>C	p.L245P	+
25	22	10	c.736C>T	p.R246C	-
26	23	10	c.736C>T	p.R246C	+
27	24	10	c.736C>T	p.R246C	+
28	25	10	c.736C>T	p.R246C	+
29		10	c.736C>T	p.R246C	+
30		10	c.736C>T	p.R246C	-
31	26	10	c.736C>T	p.R246C	+
32	27	10	c.736C>T	p.R246C	+
33	28	10	c.736C>T	p.R246C	+
34	29	10	c.736C>T	p.R246C	
35	30	10	c.736C>T	p.R246C	+
36	31	10	c.736C>T	p.R246C	-
37	32	10	c.736C>T	p.R246C	+
38	33	10	c.736C>T	p.R246C	+
39	34	10	c.736C>T	p.R246C（未確認）	
40	35		c.736C>T	p.R246C	
41	36	10	c.794G>A	p.C265Y	+
42	37	10	c.797A>G	p.Y266C	
43	38	10	c.832A>C	p.The278Pro	
44	39	10	c.1727C>A	p.S576X	+
45	40	INT16	(-)145A>C	?	
46	41	17	c.4654G>T	p.V1552F	+
47		17	c.4654G>T	p.V1552F	+
48	42	18	c.4744_4746 del.	p.T1582del	
49	43	19	c.4865C>T	p.A1622V	-
50		19	c.4865C>T	p.A1622V	-
51		19	c.4865C>T	p.A1622V	-
52	44	19	c.4870G>A	p.V1624M	+
53		19	c.4870G>A	p.V1624M	not available
54		19	c.4870G>A	p.V1624M	+
55	45	19	c.4934T>C	p.L1645S	+
56	46	int21	c.5273-5C>G; r[5273-5448del]	p.Y1758X	+

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57	47	23	C.5498A>G	p.Y1833C	-
58		23	C.5498A>G	p.Y1833C	-
59	48		C.5498A>G	p.Y1833C	
60	49	23	c.5540A>G	p.Y1847C	+
61	50	23	c.5540A>G	p.Y1847C	+
62		23	c.5540A>G	p.Y1847C	+
63	51	27	c.6052G>A	p.G2018R	
64	52	29	c.6253C>T	p.R2085C	
65	53	30	c.6392G>A	p.R2131Q	+
66	54	30	c.6392G>A	p.R2131Q	+
67	55	30	c.6406G>A	p.D2136N	-
68	56	31	c.6511A>G	p.Met2171Val	-
69		31	c.6511A>G	p.Met2171Val	-
70	57	31	c.6511A>G	p.Met2171Val	-
71	58	int31	insertion of 2kb from chr.2; splicing abnormality		-
72	59	35	c.7156C>T	Arg(CGA)2386X(TG	+
73	60	int35	c.7200+4A>G, p.L2401fs(?)		-
74	61	36	c.7423 G>A	p.Ala2475Thr	+
75	62	3'-UTR			