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Original Article

Heterozygous urinary abnormality-causing variants of *COL4A3* and *COL4A4* affect severity of autosomal recessive Alport syndrome

Tomoko Horinouchi M.D., Ph.D.[1], Tomohiko Yamamura M.D., Ph.D.[1], China Nagano M.D.[1], Nana Sakakibara M.D.[1], Shinya Ishiko M.D.[1], Yuya Aoto M.D.[1], Rini Rossanti M.D.[1], Koichi Nakanishi M.D., Ph.D.[2], Yuko Shima M.D., Ph.D.[3], Naoya Morisada M.D., Ph.D.[1], Kazumoto Iijima M.D., Ph.D.[1], Kandai Nozu M.D., Ph.D.[1]

Table S1. Patient clinical and genetic information

Patient ID	Gender	Age	ESRD age (Cr-eGFR)	Hearing loss	Ocular lesion	Gene	Mutation1			Mutation2		
							Nucleotide change	Amino acid change	Urinary/Pathological findings or diagnosis in carriers	Nucleotide change	Amino acid change	Urinary/Historical findings in carriers
94	female	17	(64.1)	-	-	COL4A3	c.4793T>G	p.Leu1598Arg	ID 143's M: OB	c.145-2A>G	Exon3(90bp)skipping	-
108	male	16	(107.0)	-	-	COL4A3	c.4028-27 A>G	Exon46(126bp) skipping	ID 108's F: OB	c.2698_2714del	p.Ile900Profs*34	-
114	male	20	(34.0)	+	-	COL4A3	c.3266G>A	p.Gly1089Asp	-	c.3266G>A	p.Gly1089Asp	-
115	female	19	(57.8)	+	-	COL4A3	c.1844dup	p.Pro616Thrfs*30	-	c.3687del	p.Gly1231Valfs*33	-
125	female	22	(138.1)	-	Retinal regeneration	COL4A3	c.2330G>A	p.Gly777Asp	ID 130's M: Pro/OB	c.4354A>T	p.Ser1452Cys	-
125-1	male	21	(8.9)	-	-	COL4A3	c.2330G>A	p.Gly777Asp	ID 130's M : Pro/OB	c.4354A>T	p.Ser1452Cys	-
125-2	male	11	(126.2)	-	-	COL4A3	c.2330G>A	p.Gly777Asp	ID 130's M : Pro/OB	c.4354A>T	p.Ser1452Cys	-
130	female	16	15	-	Perimacular fleck	COL4A3	c.2330G>A	p.Gly777Asp	ID 130's M : Pro/OB	c.4793T>G	p.Leu1598Arg	ID 143's M:OB
130-1	female	18	11			COL4A3	c.2330G>A	p.Gly777Asp	ID 130's M : Pro/OB	c.4793T>G	p.Leu1598Arg	ID 143's M :OB
137	male	20	13	+		COL4A3	c.4928G>A	p.Arg1643Lys	-	c.40_63del	p.Leu14_Leu21del	Longo et al. (ADAS)
137-1	female	27	26	-		COL4A3	c.4928G>A	p.Arg1643Lys	-	c.40_63del	p.Leu14_Leu21del	Longo et al. (ADAS)

143	female	2	(122.1)			COL4A3	c.4793T>G	p.Leu1598Arg	ID 143's M : OB	c.2125G>T	p.Gly709Term	-
155	male	36	19	+	-	COL4A3	c.4463-523 C>G	Cryptic exon(139bp)	-	c.4463-523 C>G	Cryptic exon(139bp)	-
155-1	female	33	21	+	Band-keratopathy	COL4A3	c.4463-523 C>G	Cryptic exon(139bp)	-	c.4463-523 C>G	Cryptic exon(139bp)	-
165	male	6	(167.8)	-	-	COL4A3	c.689G>A	p.Gly230Asp	ID 165's M or F: OB	c.1576-20_1576-6del	Exon25(183bp)skipping	ID 165's M or F: OB
166	female	18	18	-	-	COL4A3	c.1855G>A	p.Gly619Arg	ID 166's M: OB ID 473's M: OB	c.1060G>T	p.Gly354Term	-
167	female	21	(158.7)	+	-	COL4A3	c.4708T>C	p.Cys1570Arg	-	c.40_63del	p.Leu14_Leu21del	Longo et al. (ADAS)
168	male	19	19	-	-	COL4A3	c.1918G>A	p.Gly640Arg	ID 168's M: OB, ID 168's F: OB･TBM	c.1918G>A	p.Gly640Arg	ID 168's M: OB, ID 168's F: OB･TBM
169	male	19	(107.84)	+	-	COL4A3	c.4793T>G	p.Leu1598Arg	ID 143's M : OB	c.3752-511_3955+576del	Exon43-44del (131+73bp)	-
170	female	7	(119.52)	-	-	COL4A3	c.1354G>A	p.Gly452Arg	ID 170's F: OB	c.3821dup	p.His1275Profs*34	-
171	male	16	9	+	-	COL4A3	c.40_63del	p.Leu14_Leu21del	Longo et al. (ADAS)	c.40_63del	p.Leu14_Leu21del	Longo et al. (ADAS)
171-1	female	11	11	+	-	COL4A3	c.40_63del	p.Leu14_Leu21del	Longo et al. (ADAS)	c.40_63del	p.Leu14_Leu21del	Longo et al. (ADAS)
173	male	25	25	+	-	COL4A3	c.3464G>A	p.Gly1155Asp	-	c.4793T>G	p.Leu1598Arg	ID 143's M: OB
179	female	45	45	+	-	COL4A3	c.2863G>A	p.Gly955Arg	-	c.4793T>G	p.Leu1598Arg	ID 143's M: OB
245	male	41	(91.54)	+	-	COL4A3	c.933+1G>A	Exon16(45bp)skipping	-	c.3650_3657del	p.Pro1217Hisfs*89	-

412	male	19	(80.70)	-	-	COL4A3	c.1576G>T	p.Gly526Cys	ID 412's M: OB	c.3883-1G>C	N/A	-
415	female	17	(107.70)	+	-	COL4A3	c.4708T>C	p.Gly1507Arg	-	c.4441C>T	p.Arg1481Term	-
473	female	8	(73.93)	-	-	COL4A3	c.1855G>A	p.Gly619Arg	ID 166's M: OB ID 473's M: OB	c.4793T>G	p.Leu1598Arg	ID 143's M: OB
525	female	11	(129.84)	+	-	COL4A3	c.1994G>A	p.Gly665Asp	ID 525's M: OB	c.1216C>T	p.Arg406Term	-
570	male	45	(53.79)	-	-	COL4A3	c.3427G>A	p.Gly1143Arg	ID 570's M: OB (Any)	c.4085del	p.Pro1362Hisfs*23	ID 570's M: OB (Any)
570-1	male	47	31			COL4A3	c.3427G>A	p.Gly1143Arg	ID 570's M: OB (Any)	c.4085del	p.Pro1362Hisfs*23	ID 570's M: OB (Any)
595	female	19	18	-	-	COL4A3	c.953G>A	p.Gly318Asp	ID 595's F: OB/Pro CKD	c.4793T>G	p.Leu1598Arg	ID 143's M: OB
85	female	23	(98.35)	-	-	COL4A4	c.2510G>C	p.Gly837Ala	Kamiyoshi et al. (ADAS)	c.3151G>C	p.Gly1051Arg	ID 85's F: OB
145	female	26	(106.13)	-	-	COL4A4	c.3307G>A	p.Gly1103Arg	-	c.3307G>A	p.Gly1103Arg	-
156	female	7	(136.64)	-	-	COL4A4	c.2608G>C	p.Gly870Arg	-	c.3687dup	p.Gly1230Argfs*23	-
172	male	16	14	+	-	COL4A4	c.2084G>A	p.Gly695Asp	ID 172's M: OB	c.3612_3621del	p.Ile1205*	ID 172's F: OB･TBM
174	male	2	(100.81)	-	-	COL4A4	c.1733G>T	p.Gly578Val	-	c.4241_4254del	p.Asp1414Glyfs*14	ID 174's M: OB
204	male	11	(102.74)	-	-	COL4A4	c.2084G>A	p.Gly695Asp	ID 172's M: OB	c.4469G>C	p.Gly1490Ala	-
218	male	12	(65.77)	-	hyperopia	COL4A4	c.2878G>A	p.Gly960Arg	ID 218's F: OB	c.559-491_1460-808del insPolyT	Exon8-Exon25 del	ID 218's M: OB
257	female	18	(107.44)	-	-	COL4A4	c.3160G>C	p.Gly1054Arg	ID 257's F: OB	c.3307G>A	p.Gly1103Arg	-
270	female	47	(45.49)	-	-	COL4A4	c.203G>A	p.Gly68Glu	-	c.2437G>C	p.Gly813Arg	ID 270's F: OB
309	female	2	(126.66)	-	-	COL4A4	c.2566C>T	p.Gln856Term	ID 309's F: OB/Pro	c.3687del	p.Gly1230Valfs*58	ID 309's M: OB

468	female	4	(153.85)	-	-	COL4A4	c.1580G>T	p.Gly527Val	-	c.3160G>T	p.Gly1054Cys	ID 468's F:OB
471	female	41	(29.53)	+	-	COL4A4	c.4953G>A	p.Trp1651Term	-	c.2930del	p.Pro977Leufs*61	-
476	female	21	(124.99)	-	-	COL4A4	c.2510G>C	p.Gly837Ala	Kamiyoshi et al. (ADAS)	c.4817G>A	p.Gly1606Glu	Baek et al.
601	female	22	(82.08)	-	-	COL4A4	c.3262G>C	p.Gly1088Arg	ID 601's F: OB	c.3307G>A	p.Gly1103Arg	-
738	male	37	N/A	+	-	COL4A4	c.2617G>A	p.Gly873Arg	ID 738's M: OB・60yr ESRD	c.594+5G>A	N/A	-
738-1	female	33	27	+	-	COL4A4	c.2617G>A	p.Gly873Arg	ID 738's M: OB・60yr ESRD	c.594+5G>A	N/A	-
741	female	7	(132.27)	-	-	COL4A4	c.1795G>C	p.Gly599Arg	ID 741's M: OB ID 741's F: OB	c.1795G>C	p.Gly599Arg	ID 741's M: OB ID 741's F: OB

ESRD: End-stage renal disease
CKD: Chronic kidney disease
OB: Occult blood
Pro: Proteinuria
TBM: Thin basement membrane
ADAS: Autosomal dominant Alport syndrome
M: Mother
F: Father
Kamiyoshi et al.: This mutation was reported by Kamiyoshi et al. (1)
Longo et al.: This mutation was reported by Longo et al. (2)

Baek et al.: This mutation was reported by Baek et al. (3)

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