MolecularMedicine

Supplemental Data

Mutation Spectrum of *STAR* and the Founder Effect of p.Q258* in Korean Patients with Congenital Lipoid Adrenal Hyperplasia

Eungu Kang,[1] Yoon-Myung Kim,[1] Gu-Hwan Kim,[2] Beom Hee Lee,[1] Han-Wook Yoo,[1] and Jin-Ho Choi[1]

Online address: http://www.molmed.org

Feinstein Institute
for Medical Research
Northwell Health™

Supplementary Table S1. Karyotype and mutations in the *STAR* gene of 45 patients from 42 families.

Family number	Patient number	Sex of rearing	Karyotype	*STAR* gene	
				Allele 1	Allele 2
1	1*	F	46,XX	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
2	2*	F	46,XX	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
3	3	F	46,XX	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
4	4	F	46,XX	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
5	5*	F	46,XX	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
6	6	F	46,XX	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
7	7	F	46,XX	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
8	8	F	46,XX	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
9	9	F	46,XX	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
10	10	F	46,XX	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
10	11	F	46,XX	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
11	12*	F	46,XX	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
12	13*	F	46.XY	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
13	14	F	46,XY	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
14	15	F	46,XY	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
15	16	F	46,XY	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
16	17	F	46,XY	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
16	18	F	46,XY	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
17	19	F	46,XY	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
18	20	F	46,XY	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
19	21	F	46,XY	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
20	22	F	46,XY	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
21	23	F	46,XY	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
22	24	F	46,XY	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
23	25	F	46,XY	c.772C>T (p.Q258*)	c.545G>A (p.R182H)
24	26	F	46,XY	c.772C>T (p.Q258*)	c.653C>T
25	27	F	46,XY	c.745-6_810del	c.653C>T
26	28	F	46,XY	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
27	29+	F	46,XX	c.772C>T (p.Q258*)	c.559G>A (p.V187M)
28	30	F	46,XY	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
29	31	F	46,XX	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)

Continued on next page

Supplementary Table S1. ***Continued.***

Family number	Patient number	Sex of rearing	Karyotype	*STAR* gene	
				Allele 1	Allele 2
30	32	F	46,XX	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
31	33	F	46,XY	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
32	34[+]	M	46,XY	c.772C>T (p.Q258*)	c.815G>A (p.R272H)
33	35*	F	46,XY	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
34	36	F	46,XX	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
35	37*,[+]	F	46,XX	c.772C>T (p.Q258*)	c.815G>A (p.R272H)
36	38	F	46,XY	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
37	39	F	46,XX	c.772C>T (p.Q258*)	c.543C>T (p.R182C)
37	40	F	46,XX	c.772C>T (p.Q258*)	c.543C>T (p.R182C)
38	41	F	46,XX	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
39	42	F	46,XX	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
40	43*,[+]	F	46,XX	c.772C>T (p.Q258*)	c.559G>A (p.V187M)
41	44*	F	46,XY	c.772C>T (p.Q258*)	c.772C>T (p.Q258*)
42	45*	F	46,XX	c.772C>T (p.Q258*)	c.407del (p.E136Gfs*50)

*, indicates the patients who were included in this study; [+], indicates nonclassical late onset CLAH patients

Supplementary Table S2. Haplotype analysis of individuals with mutations in the *STAR* gene.

		Physical position on chromosome 8 (hg19)	Patient 1	Patient 2	Patient 3	Patient 4	Patient 5	Patient 6	Patient 7	Patient 8	Patient 9	Patient 10
8	rs6986132	37815009	C C	C C	C C	C C	C C	C C	C C	C C	C C	C C
8	rs802170	37815121	T T	T T	T T	T T	T T	T T	T T	T T	A T	T T
8	rs1892818	37817175	G G	G G	G G	G G	G G	G G	G G	G G	G G	G G
8	rs4998	37821486	C C	C C	C C	C C	G C	C C	C C	C C	G C	G C
8	rs4994	37823798	C C	C C	C C	C C	T C	C C	C C	C C	T C	T C
8	rs2298423	37834312	C C	C C	C C	C C	A C	C C	C C	C C	A C	A C
8	rs73674110	37845945	A A	A A	A A	A A	A A	A A	A A	A A	A A	A A
8	rs2186458	37854599	G G	G G	G G	G G	G G	G G	G G	G G	T G	G G
8	rs58656606	37864599	A A	A A	A A	A A	C A	A A	A A	A A	C A	C A
8	rs6605630	37869772	G G	G G	G G	G G	A G	G G	G G	G G	A G	A G
8	rs73674118	37870395	G G	G G	G G	G G	G G	G G	G G	G G	G G	C G
8	rs59709543	37870582	G G	G G	G G	G G	A G	G G	G G	G G	A G	A G
8	rs28813686	37883438	C C	C C	C C	C C	C C	C C	C C	C C	C C	C C
8	rs28797500	37884310	C C	C C	C C	C C	C C	C C	C C	C C	C C	C C
8	rs3813521	37887422	C C	C C	C C	C C	C C	C C	C C	C C	C C	C C
8	rs28368989	37897855	T T	T T	T T	T T	T T	T T	T T	T T	T T	T T
8	rs28377666	37899702	G G	G G	G G	G G	G G	G G	G G	G G	G G	G G
8	rs6605631	37902453	A A	A A	A A	A A	A A	A A	A A	A A	A A	A A
8	rs28570677	37908814	G G	G G	G G	G G	G G	G G	G G	G G	T G	T G
8	rs7816482	37923459	T T	T T	T T	T T	C T	T T	T T	T T	T T	T T
8	rs78758900	37930661	G G	G G	G G	G G	A G	G G	G G	G G	G G	G G
8	rs28877647	37946551	G G	G G	G G	G G	G G	G G	G G	G G	G G	G G
8	rs2073351	37963239	G G	G G	G G	G G	G G	G G	G G	G G	A G	A G
8	rs2843743	37976368	G G	G G	G G	G G	G G	G G	G G	G G	A G	A G
8	rs2517388	37977732	T T	T T	T T	T T	T T	T T	T T	T T	G T	G T
8	rs2720044	37980587	A A	A A	A A	A A	A A	A A	A A	A A	A A	A A
8	rs2843740	37985897	G G	G G	G G	G G	A G	G G	G G	G G	A G	A G
8	rs2517397	37992515	T T	T T	T T	T T	C T	T T	T T	T T	T T	T T
8	rs63215562	37999998	T T	T T	T T	T T	T T	T T	T T	T T	C T	C T
8	rs28361934	38000804	T T	T T	T T	T T	T T	T T	T T	T T	T T	T T
8	rs35045158	38001335	C C	C C	C C	C C	C C	C C	C C	C C	C C	C C
8	rs34575008	38001561	A A	A A	A A	A A	A A	A A	A A	A A	A A	A A
8	rs16887217	38004425	T T	T T	T T	T T	T T	T T	T T	T T	T T	T T
8	rs2720049	38004911	G G	G G	G G	G G	G G	G G	G G	G G	G G	G G
8	rs7819423	38004975	A A	A A	A A	A A	A A	A A	A A	A A	A A	A A
8	rs72552290	38006184	C C	C C	C C	C C	C C	C C	C C	C C	C C	C C
8	rs6474491	38013848	T T	T T	T T	T T	T T	T T	T T	T T	T T	T T

*Shared haplotype of each patient from paternal or maternal alleles are marked by gray color.

The father of patient 5, the mother of patient 9, and the mother of patient 10 are carriers of the p.V187M, p.R272H, and p.E136Gfs*50, respectively. The other parents are carriers of p.Q258*.