

Supplementary Table 1. Clinical characteristic of patients with C1-INH-HAE and disease-associated variants identified by Sanger sequencing genotyping and MLPA analysis

No. family	No. of case	Sex	Age	aC1INH (g/l)	fC1INH (%)	aC4 (g/l)	C1-INH-HAE type	Family history (yes/no)	Score 0-3	Age of onset/location of symptom
A. Sanger sequencing										
I	1	W	59	0.04	0	0.034	I	No	3	4 Abdomen
	2	W	30	0.04	6.8	0.043	I	Yes	2	9 Abdomen
II	3	W	56	0.03	3.2	0.014	I	No	2	14 Abdomen
	4	M	36	0.09	22.9	0.057	I	Yes	2	6 Arm
	5	W	31	0.046	8.5	0.0254	I	Yes	3	3 Face
III	6	M	37	0.1	13.8	0.11	I	No	3	6 Larynx
IV	7	W	23	0.72	37.1	0.11	II	Yes	2	11 Hand
	8	W	29	0.25	19.9	0.1	II	Yes	2	12 Hand
VI	10	W	69	0.09	37.1	0.05	I	No	3	29 Face
VIII	15	M	53	0.21	36	0.066	II	No	2	21 Face
	16	W	30	0.2	27.8	0.07	II	Yes	3	2 Abdomen
IX	17	W	38	0.34	18	0.02	II	No	2	6 Abdomen
X	18	W	22	0.07	7.3	0.04	I	No	1	10 Abdomen
XI	19	W	35	0.04	28.1	0.09	I	No	3	35 Face
XII	20	W	52	0.05	28.1	0.03	I	Yes	2	13 Abdomen

	21	W	55	0.03	18.6	0	I	Yes	3	12 Abdomen
XVI	25	M	58	0.06	62	0.08	I	Yes	3	15 Abdomen
XIX	28	W	47	0.46	12.7	0.045	II	No	2	18 Hand
XX	29	M	44	0.07	62.8	0.07	I	Yes	3	5 Hand
XXII	32	W	75	0.03	26	0.02	I	Yes	3	13 Larynx
XXIII	33	W	51	0.036	15.3	0.02	I	No	2	17 Abdomen
XXIV	34	W	41	0.06	23.8	0.05	I	Yes	3	3 Lanrynx
XXV	35	M	37	0.07	16	0.06	I	No	2	20 Hand
XXVI	36	W	33	0.37	11.8	0.02	II	No	3	10 Face
XXVIII	38	W	43	0.08	25	0.07	I	Yes	3	12 Abdomen
XXIX	39	M	41	0.03	0	0.04	I	Yes	2	7 Face
XXX	40	W	29	0.05	9.3	0.049	I	No	3	3 Hand
XXXI	41	W	40	0.048	11.8	0.12	I	Yes	2	4 Abdomen
B. MLPA analysis										
VII	11	W	61	0.06	10.6	0.034	I	No	3	22 Abdomen
	12	W	33	0.13	34.6	0.04	I	Yes	2	10 Face

	13	M	4	0.07	65	0.05	I	Yes	0	Asymptomatic
	14	W	29	0.03	26.5	0.066	I	Yes	2	12 Abdomen
XXI	30	W	58	0.06	22.4	0.05	I	No	3	14 Face
	31	W	26	0.056	3.2	0.025	I	Yes	3	11 Lanrynx
Together		26 W 8 M		I – 0.06 II – 0.4	I – 21.8 II – 23.3	I – 0.05 II – 0.06	I – 27 II – 7	Yes 19 No 15	0-3 (Median 2.5)	2-35 (Mean 13.1)

W – women, M – men, aC1INH – serum level of C1INH (normal range 0.2-0.39 g/l), fC1INH – the activity of C1INH in plasma (normal range 70-130%), aC4 – serum level of C4 (normal range 0.1-0.4 g/l)