

Table 3. Cystinuria-specific mutated alleles identified in the 164 probands studied of the International Cystinuria Consortium (ICC) database.

One M467T allele (*SLC3A1*) and one c.789+2T>C allele (*SLC7A9*) are not included because they are the third mutated allele in patients BB(A) and AA(B) respectively. *) Two mutations in the same chromosome. #) Renamed *SLC7A9* mutations following The Human Genome Variation Society nomenclature (www.genomic.unimelb.edu.au/mdi/mutnomen/) (e.g., for the common mutation c.800-801insA, also named c.799-800insA, the new name is c.614dupA). \$) Two c.604+1G>A alleles have been considered in one patient even though hemizygosity due to deletion in the other *SLC7A9* allele is not discharged. &) *SLC3A1* deletions with unidentified boundaries. (n), number of independent alleles. F, frequency. Previous identified mutations in the ICC database shown in this table have been published elsewhere ^{2,7,8,15,16,17,18}.

Type A mutations (<i>SLC3A1</i>)				Type B mutations (<i>SLC7A9</i>)			
Nucleotide change	Amino acid change/ Predicted consequence	(n)	F	Nucleotide change	Amino acid change/ Predicted consequence	(n)	F
c.410A>G	D137G	1	0,008	c.977+3A>T#	A290fsX45	1	0,006
c.1699_1700 delAG	R567fsX8	1	0,008	c.1265_1266 delTG	L424fsX62	1	0,006
c.664delT	W222fsX3	1	0,008	c.1399+1G>T	447fsX22	1	0,006
c.542G>A	R181Q	1	0,008	c.475_477dupGCC#	A158dup	1	0,006
c.1702G>A	G568L	1	0,008	c.579_581dupCAT#	I193dup	1	0,006
c.1998C>G	C666W	1	0,008	c.151delT	S51fsX38	1	0,006
c.1364C>T	S455L	1	0,008	c.368_369 delCG#	T123fsX84	1	0,006
c.1035G>A	D338fsX4	1	0,008	c.501_504 delACTG#	L168fsX19	1	0,006
c.1043A>C	H348P	1	0,008	c.630_633 delTTTC#	F211fsX51	1	0,006
c.446T>A	L149Q	1	0,008	c.813delC#	N271fsX6	1	0,006
c.368T>G	M123R	1	0,008	c.376G>A#	A126T	1	0,006
c.1690C>T	L564F	1	0,008	c.947C>T	A316V	1	0,006
c.1230C>A	N410K	1	0,008	c.1144G>A#	A382T	1	0,006
c.1865T>C, c.1520C>T	L622X, S507L*	1	0,008	c.171C>T	N58_G79 del22	1	0,006
c.566C>T	T189M	1	0,008	c.586C>T	L196_G202 delfsX5	1	0,006

c.1367G>A	R456H	1	0,008	c.712_716 delCTCAA#	N239fsX6	1	0,006
c.1966_1968 delCTT	L656del	1	0,008	c.1224+4166_ c.1399+119del(4972)	K386fsX78	1	0,006
c.2019C>G	C673W	1	0,008	c.1224+4166_ c.1399+119dup(4972)	V409fsX9	1	0,006
c.2033T>C	L678P	1	0,008	c.187G>A#	G63R	1	0,006
c.1355G>A	R452Q	1	0,008	c.131T>C#	I44T	1	0,006
c.1943T>C	F648S	1	0,008	c.1400-3C>G	447fsX22	1	0,006
c.1618_ 2055del	V540_ C685del	1	0,008	c.847C>T	L283F	1	0,006
c.431_2055 del	G144_ C685del	1	0,008	c.155C>T#	P52L	1	0,006
c.766-2A>G	L256_ K297del	1	0,008	c.184G>A	V62M	1	0,006
c.1093C>T, c.1955C>G	R365W, T652R*	1	0,008	c.206G>T#	W69L	1	0,006
c.371A>G	Y124C	1	0,008	c.206G>A#	W69X	1	0,006
c.1383T>G	Y461X	1	0,008	c.695A>G	Y232C	1	0,006
c.1744T>C	Y582H	1	0,008	c.235+3_ c.235+293del	L30fsX9	1	0,006
c.1012-23C>G	D338_Y378 delfsX5	1	0,008	c.1060G>A#	A354T	1	0,006
c.1221dupT	P408fsX18	1	0,008	c.562G>A	V188M	1	0,006
c.503C>A	S168X	1	0,008	c.604+1G>A \$ #	L160X	2	0,013
Del 3'	to be defined ^{&}	1	0,008	c.604+2T>C	L160X	2	0,013
Del 5'	to be defined ^{&}	2	0,016	c.209C>T#	A70V	2	0,013
c.1749delA	T583fsX14	2	0,016	c.583G>A#	G195R	2	0,013
c.388T>C	S130P	2	0,016	c.544G>A#, c.1060G>A#	A182T, A354T*	2	0,013
c.1447G>T	E483X	2	0,016	c.775G>A#	G259R	2	0,013
c.1366C>T	R456C	2	0,016	c.688T>C, 28_30del AGA#	W230R, R10del ^o	3	0,019
c.892-1332del	E298_ M444del	2	0,016	c.730_732 delGAA#	E244del	3	0,019
c.266T>C	L89P	2	0,016	c.605-3C>A	I203fsX27	4	0,025
c.1084C>T	R362C	2	0,016	c.368G>A#	T123M	6	0,038
c.891+4A>G	L256_K297del	2	0,016	c.544G>A#	A182T	8	0,051
c.1640C>G	S547W	2	0,016	c.614dupA#	L160fsX1	11	0,070
c.451T>A	Y151N	2	0,016	c.508G>A#	V170M	18	0,115
c.1093C>T	R365W	2	0,016	c.997C>T#	R333W	19	0,121
c.1322C>G	P441R	3	0,024	c.311G>A#	G105R	43	0,274
c.1353C>T	R452W	3	0,024		Total	157	1
c.1400T>A	M467K	3	0,024				
c.765+1G>T	L256_K297del	4	0,032				
c.808C>T	R270X	5	0,040				
c.163delC	Q55fsX51	5	0,040				

DupI4-I9	E298_ D539dup	5	0,040
c.647C>T	T216M	8	0,064
c.1400T>C	M467T	33	0,264
Total		125	1

