

**Table S1** Summary of the data of Chinese children with ALGS in all the reported research articles with detailed information

Patient No.	Mutant gene	Nucleotide changes	Interlobular bile duct paucity	Cholestasis	Cardiac abnormalities	Skeletal abnormalities	Characteristic face	Posterior embryotoxon	Kidney abnormalities
1	JAG1	c.1868delG	+	+	+	+	+	+	+
2	JAG1	c.439C>T	NA	+	+	+	+	+	+
3	JAG1	c.866delG	+	+	+	+	+	+	-
4	JAG1	c.1323_1326delCTGG	+	+	+	+	+	+	-
5	JAG1	c.1771_1775delGTGCG1insT	+	+	+	-	+	+	+
6	JAG1	c.2628G>A	+	+	+	+	+	+	-
7	JAG1	c.439+2dupT	+	+	+	+	+	+	-
8	JAG1	c.550C>T	-	+	+	+	+	+	-
9	JAG1	c.2572+1G>T	+	+	+	+	+	+	-
10	JAG1	c.980_989delGGTATTCAGG	NA	+	+	+	+	+	-
11	JAG1	c.1007delC	NA	+	+	+	+	+	-
12	JAG1	c.2230delC	NA	+	+	+	+	-	+
13	JAG1	c.2502delC	NA	+	+	+	+	+	-
14	JAG1	c.703C>T	NA	+	+	+	+	+	-
15	JAG1	c.2473C>T	NA	+	+	+	+	-	+
16	JAG1	c.2698C>T	NA	+	+	+	+	+	-
17	JAG1	c.3140C>A	NA	+	+	+	+	+	-
18	JAG1	entire gene deletion	NA	+	+	+	+	-	+
19	JAG1	c.1148_1149delGT	+	+	+	+	+	NA	+
20	JAG1	c.3099_3100delCA	NA	+	+	+	+	+	NA
21	JAG1	c.2230C>T	+	+	+	+	+	+	NA
22	JAG1	c.1349-10_1353del TATTTTTTAGATATT	-	+	+	+	+	+	NA
23	JAG1	c.1156G>A	NA	+	+	+	+	+	NA
24	JAG1	c.410delA	+	+	+	-	+	+	-
25	JAG1	c.693_694delAG	+	+	-	+	+	-	+
26	JAG1	c.1468G>T	+	+	+	+	+	-	-
27	JAG1	c.439+1G>A	-	+	+	+	+	-	-
28	JAG1	Ex.1-5 deletion	-	+	+	+	+	-	-
29	JAG1	c.755+2T>G	NA	+	+	+	+	-	-
30	JAG1	c.1382_1383delAC	NA	+	+	+	+	-	-
31	JAG1	c.1842delC	NA	+	+	-	+	-	+
32	JAG1	c.1859delG	NA	+	+	+	+	-	-
33	JAG1	c.2909_2913delTGTC	NA	+	+	+	-	+	-

**Table S1** (continued)

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Patient No.	Mutant gene	Nucleotide changes	Interlobular bile duct paucity	Cholestasis	Cardiac abnormalities	Skeletal abnormalities	Characteristic face	Posterior embryotoxon	Kidney abnormalities
34	JAG1	c.2070_2073dupCTGT	NA	+	+	-	+	+	-
35	JAG1	c.3088_3089insG	NA	+	+	+	+	-	-
36	JAG1	c.439C>T	+	+	+	+	+	-	NA
37	JAG1	c.766G>T	-	+	+	-	+	+	NA
38	JAG1	c.2026T>G	NA	+	+	+	+	-	NA
39	JAG1	c.3008_3020insAGCCTTCCCCTTC	NA	+	+	+	+	-	NA
40	JAG1	c.2225_2226delTA	NA	+	+	+	+	NA	-
41	JAG1	c.702C>A	NA	+	+	+	+	NA	-
42	JAG1	c.238A>G	NA	+	+	+	+	NA	-
43	JAG1	entire gene deletion	+	+	+	+	+	NA	-
44	JAG1	entire gene deletion	NA	+	+	+	+	-	NA
45	JAG1	Ex.2-26 deletion	-	+	+	+	+	-	NA
46	JAG1	c.2791_2792insA	+	+	+	+	+	NA	NA
47	JAG1	c.819delC	+	+	+	+	-	-	-
48	JAG1	c.1899_1900delTG	+	+	+	+	-	NA	NA
49	JAG1	c.1499delG	NA	+	+	+	-	-	-
50	JAG1	c.2314delG	NA	+	+	+	-	-	-
51	JAG1	c.3244_3256delATCTGTTGCTTGG	NA	+	+	+	-	-	-
52	JAG1	c.2990C>A	NA	+	+	-	-	+	-
53	JAG1	c.3031G>T	NA	+	+	-	+	-	-
54	JAG1	c.2345-2A>G	NA	+	-	+	+	-	-
55	JAG1	c.2071T>A	NA	+	-	+	+	-	-
56	JAG1	c.897delC	NA	+	+	-	+	-	NA
57	JAG1	c.3194_3195delGA	NA	+	-	+	+	-	NA
58	JAG1	c.1281_1282insT	NA	+	+	+	-	-	NA
59	JAG1	c.2990C>A	NA	+	+	-	+	-	NA
60	JAG1	c.1382_1383delAC	NA	+	+	-	+	NA	-
61	JAG1	c.1931delG	NA	+	+	-	+	NA	-
62	JAG1	c.2287_2288insAACG	NA	+	-	+	+	NA	-
63	JAG1	c.1885+3_1885+4insGT	NA	+	+	-	+	NA	-
64	JAG1	c.65delG	-	+	+	+	-	NA	NA
65	JAG1	c.1118delC	NA	+	+	+	-	NA	NA
66	JAG1	c.2698C>T	NA	+	+	+	NA	NA	NA

Table S1 (continued)

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Patient No.	Mutant gene	Nucleotide changes	Interlobular bile duct paucity	Cholestasis	Cardiac abnormalities	Skeletal abnormalities	Characteristic face	Posterior embryotoxon	Kidney abnormalities
67	JAG1	c.703C>T	-	+	+	-	-	-	-
68	JAG1	c.826delT	NA	+	-	+	-	-	NA
69	JAG1	c.439+1G>A	NA	+	-	-	+	NA	-
70	JAG1	c.3006C>A	NA	+	-	+	-	NA	NA
71	JAG1	c.3178C>T	+	+	+	-	-	NA	+
72	JAG1	c.2003T>C	+	+	-	+	-	NA	+
73	JAG1	c.1947_1948insTCCT	NA	+	+	-	+	NA	+
74	JAG1	c.359T>A	+	+	-	+	+	NA	+
75	JAG1	c.2698C>T	NA	+	+	-	+	NA	+
76	JAG1	c.526G>A	-	+	+	+	-	NA	+
77	JAG1	c.2473C>T	NA	+	-	+	+	NA	+
78	JAG1	c.133G>T	-	+	+	+	-	NA	-
79	JAG1	c.771G>A	NA	+	+	+	-	NA	+
80	JAG1	c.1227C>A	NA	+	+	-	+	NA	+
81	JAG1	IVS6+1 G>A	+	+	+	-	-	NA	+
82	JAG1	c.1511A>G	-	+	-	+	-	NA	-
83	JAG1	c.1511A>G	NA	+	+	-	-	NA	-
84	JAG1	c.1156G>A	NA	+	+	-	-	NA	-
85	JAG1	c.1563_1564delCT	NA	+	+	-	+	NA	+
86	NOTCH2	c.5983_5984delTT	-	-	+	-	+	NA	-
87	JAG1	c.2230 C>T	NA	+	+	+	+	+	-
88	JAG1	c.439+1G>A	NA	+	+	+	+	+	-
89	JAG1	c.2419delG	NA	+	+	+	+	-	-
90	JAG1	c.2230C>T	NA	+	-	+	+	+	+
91	JAG1	c.1976G>A	NA	+	+	-	+	+	-
92	JAG1	3.0 Mb deletion	NA	+	+	-	+	-	-
93	JAG1	9.24 Mb deletion	NA	+	+	+	-	+	-
94	JAG1	c.3196dupA	NA	+	+	+	-	+	+
95	JAG1	c.1977G>A	NA	+	+	+	+	+	-
96	JAG1	c.1106_1107delCC	NA	+	-	-	+	+	-
97	JAG1	c.1106_1107delCC	NA	+	-	-	+	+	-
98	NOTCH2	c.3928C>T	NA	+	-	+	-	-	+
99	JAG1	c.2698C>T	NA	+	+	+	+	-	-

Table S1 (continued)

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Patient No.	Mutant gene	Nucleotide changes	Interlobular bile duct paucity	Cholestasis	Cardiac abnormalities	Skeletal abnormalities	Characteristic face	Posterior embryotoxon	Kidney abnormalities
100	<i>JAG1</i>	c.1464delC	+	+	+	-	+	-	+
101	<i>JAG1</i>	c.3094_3095delAC	NA	-	-	+	+	-	+
102	<i>JAG1</i>	c.1853delA	NA	+	+	-	+	-	-
103	<i>JAG1</i>	c.2080A>T	NA	+	+	-	+	-	-
104	<i>JAG1</i>	c.1395+3A>G	-	+	+	-	-	-	-
105	<i>JAG1</i>	c.2698C>T	NA	-	-	-	+	-	-
106	<i>JAG1</i>	c.1437T>A	NA	+	-	-	+	-	-
107	<i>JAG1</i>	c.960T> G	NA	+	+	+	-	-	-
108	<i>NOTCH2</i> <i>NOTCH2</i>	c.6007C>T c.6157C>T	NA	+	+	-	-	NA	-
109	<i>JAG1</i>	c. 133G>T	NA	+	-	-	+	-	-
110	<i>JAG1</i>	c.2370delC	NA	+	+	+	+	+	-
111	<i>JAG1</i> <i>NOTCH2</i>	c.3286C>T c.6961dupG	NA	+	-	-	+	-	-
112	<i>JAG1</i>	c.3045delC	NA	+	-	+	+	+	-
113	<i>JAG1</i>	c.3391G>A	NA	+	-	-	+	-	-
114	<i>JAG1</i>	c.703C>T	NA	+	+	-	+	-	-
115	<i>JAG1</i>	c. 1213delA	NA	+	+	+	+	+	-
116	<i>NOTCH2</i>	c.710G>A	NA	+	+	+	-	-	-
117	<i>JAG1</i>	c.1856_ 1857delAA	NA	+	-	+	+	-	-
118	<i>JAG1</i> <i>NOTCH2</i>	c.625C>A c.3779G>A	NA	+	-	+	-	-	-
119	<i>JAG1</i>	c.2000-2A>C	NA	+	+	+	+	NA	-
120	<i>JAG1</i>	c. 1270dupG	NA	+	-	+	+	+	-
121	<i>JAG1</i>	c. 1156G>A	NA	+	+	+	+	-	+
122	<i>NOTCH2</i>	c.518G>T	NA	+	-	-	+	-	-
123	<i>JAG1</i>	c.2698C>T	NA	+	+	+	+	-	-
124	<i>JAG1</i>	c. 1741dupG	NA	+	+	+	+	-	-
125	<i>JAG1</i>	c. 1270dupG	NA	+	+	-	+	+	-
126	<i>JAG1</i>	c.232_233dup	+	+	+	-	+	+	NA
127	<i>JAG1</i>	c.439+1G>A	+	+	+	-	+	-	NA
128	<i>JAG1</i>	c.2323G >T	-	+	+	+	+	-	NA
129	<i>JAG1</i>	c.3000del	+	+	+	+	+	+	NA
130	<i>JAG1</i>	c.550C>T	+	+	+	+	+	-	NA

Table S1 (continued)

**Table S1** (continued)

Patient No.	Mutant gene	Nucleotide changes	Interlobular bile duct paucity	Cholestasis	Cardiac abnormalities	Skeletal abnormalities	Characteristic face	Posterior embryotoxon	Kidney abnormalities
131	<i>JAG1</i>	c.1308C>A	+	+	NA	+	-	NA	NA
132	<i>JAG1</i>	c.3002_3003del	-	+	+	+	-	NA	NA
133	<i>JAG1</i>	c.548T>A	+	+	+	+	+	-	NA
134	<i>JAG1</i>	c.2122_2125del	+	+	+	+	+	+	NA
135	<i>JAG1</i>	c.1485_1486del	-	+	-	+	+	NA	NA
136	<i>JAG1</i>	chr20:10018942–17,550,915	+	-	+	+	+	NA	NA
137	<i>JAG1</i>	c.162del	+	+	-	-	+	+	NA
138	<i>JAG1</i>	c.100C>T	+	+	+	-	+	NA	NA

+ positive, - negative, NA, not available. Patient 1–70 from reference 14; Patient 71–86 from reference 16; Patient 87–97 from reference 15; Patient 98–107 from reference 19; Patient 108–125 from reference 18; Patient 126–138 from reference 21.