

Table SI. Genetic information of reported neonatal cases in china (including main laboratory data).

Entry	Gene	Substitution	RBC, x10 ¹² /l	HGB, g/dl	MCV, fl	MCH, pg	MCHC, g/l	Ret, %	TBil, μmol/l	Ref.
1	ANK1	c.2T>G/p.M1 R	2.71	7.1	NA	NA	NA	5.61	NA	(1)
2	ANK1	c.1814_1818d elCTTTG/p.L 606Hfs*13	NA	NA	NA	NA	NA	NA	NA	(1)
3	ANK1	c.3115+2T>G	NA	NA	NA	NA	NA	NA	NA	(1)
4	ANK1	c.3275delA/p. Q1092Rfs*13	NA	NA	NA	NA	NA	NA	NA	(1)
5	ANK1	c.3554G>A/p. W1185*	NA	NA	NA	NA	NA	NA	NA	(1)
6	ANK1	c.4000C>T/p. R1334*	NA	NA	NA	NA	NA	NA	NA	(1)
7	ANK1	c.4153C>T/p. R1385*	NA	NA	NA	NA	NA	NA	NA	(1)
8	ANK1	c.5455C>T/p. Q1819*	NA	NA	NA	NA	NA	NA	NA	(1)
9	SPTB	c.2863C>T/p. R955*	NA	NA	NA	NA	NA	NA	NA	(1)
10	SLC4 A1	c.2423G>C/p. R808P	NA	NA	NA	NA	NA	NA	NA	(1)
11	ANK1	c.319C>T/p.Q 107*	3.19	109	96.6	34.2	354	8.44	510.8	(2)
12	ANK1	c.3813_3823d el/p.Q1272Lfs *100	NA	NA	NA	NA	NA	NA	179	(2)
13	ANK1	c.3847delA/p. R1283Gfs*3	NA	NA	NA	NA	NA	NA	55.7	(2)
14	SPTB	c.1912C>T/p. R638*	1.92	59	84.2	29.2	347	7.63	202	(2)
15	SPTB	c.3168dupG/p. L1057Afs*16	NA	NA	NA	NA	NA	NA	272.3	(2)
16	SPTB	c.3448dupT/p. W1150Lfs*32	NA	NA	NA	NA	NA	NA	146.4	(2)
17	SPTB	c.5266C>T/p. R1756*	1.95	60	86.2	30.8	357	3.56	484.7	(2)
18	ANK1	c.T290G/p.L9 7R	2.22	60	76	27	355	7.57	53.53	(3)
19	SPTB	c.3737delA/p. K1246fs	NA	51	81.5	29.3	384	12.3	553	(4)
20	ANK1	c.1000delA/p. I334Sfs*6	3.93	138	101.8	35.1	345	4.3	233.6	(5)
21	ANK1	c.834_833ins C/p.C278Wfs *78	3.29	112	NA	NA	NA	5.5	535.4	(6)
22	SPTB	c.5798+1G>A	1.9	60	NA	NA	NA	NA	114.7	(7)
23	ANK1	c.2960+2T>G	7.6	NA	91.9	31.8	356	3.09	NA	(8)
24	SPTB	c.5165_c.5166 delTT/p.F172 2*	NA	145	98.6	33.3	337	1.15	94	(9)

RBC, red blood cell count; HGB, hemoglobin; MCV, mean corpuscular volume; MCHC, mean corpuscular HGB concentration; Ret, reticulocyte; TBil, total bilirubin; Ref., reference; NA, not available.

Table SII. Genetic information of reported non-neonatal cases in China.

No.	Gene	Substitution	Ref.
1	ANK1	c.2T>G/p.M1R	(10)
2	ANK1	c.328+2A>G	(10)
3	ANK1	c.541G > C/p.A181P	(10)
4	ANK1	c.740C > T/p.S247F	(10)
5	ANK1	c.781C > T/p.R261W	(10)
6	ANK1	c.810+5G>A	(10)
7	ANK1	c.811G > T/p.D271Y	(10)
8	ANK1	c.856C> T/p.R286*	(10)
9	ANK1	c.990_991delGA/p.E330Dfs*25	(10)
10	ANK1	c.1717delC/p.L573Cfs*64	(10)
11	ANK1	c.2524G> T/p.E842Cfs*	(10)
12	ANK1	c.2848A> C/p.T950P	(10)
13	ANK1	c.3116-1G>C	(10)
14	ANK1	c.3178C> A/p.P1060T	(10)
15	ANK1	c.3464G>A/p.W1155*	(10)
16	ANK1	c.3553T> C/p.W1185R	(10)
17	ANK1	c.5022dupA/p.G1675Rfs*66	(10)
18	ANK1	c.5044C>T/p.R1682*	(10)
19	SPTB	c.759delC/p.E254Kfs*27	(10)
20	SPTB	c.850delG/p.K107Rfs*33	(10)
21	SPTB	c.1540C>T/p.E254Kfs*27	(10)
22	SPTB	c.1628G>A/p.E254Kfs*27	(10)
23	SPTB	c.1912C>T/p.R638*	(10)
24	SPTB	c.2413C>T/p.Q805*	(10)
25	SPTB	c.2488_2491delTACC/p.Y830Nfs*67	(10)
26	SPTB	c.2566G>A/p.E856K	(10)
27	SPTB	c.2907G>A/p.W969*	(10)
28	SPTB	c.4266+4C>G	(10)
29	SPTB	c.4267C>T/p.R1423*	(10)
30	SPTB	c.4967C>T/p.P1656L	(10)
31	SPTB	c.5587C>T/p.Q1863*	(10)
32	SPTB	c.5844_5855delCAATGCAGAGAT/p.1948_1951delINAE	(10)
33	SPTB	c.6238C>T/p.Q2080*	(10)
34	SLC4A1	c.808G>T/p.G270*	(10)

35	SLC4A1	c.1179C> A/p.Y393*	(10)
36	SLC4A1	c.2102G>A/p.G701D	(10)
37	SLC4A1	c.2287G>A/p.G763R	(10)
38	SLC4A1	c.2510C>T/p.T837M	(10)
39	ANK1	c.725_728dupCACT/p.H244Tfs*113	(1)
40	ANK1	c.856C>T/p.R286*	(1)
41	ANK1	c.1109dupA/p.N370Kfs*101	(1)
42	ANK1	c.1867C>T/p.Gln623*	(1)
43	ANK1	c.2735+1G>T	(1)
44	ANK1	c.2803C>T/p.R935*	(1)
45	ANK1	c.3302_3305delinsGAGTGCCGGAGAATGCCG	(1)
46	ANK1	c.3464G>A/p.W1155*	(1)
47	ANK1	c.4306C>T/p.R1436*	(1)
48	SPTB	c.1A>C/?	(1)
49	SPTB	c.173dupA/p.T59Dfs*113	(1)
50	SPTB	c.647G>A/p.R216Q	(1)
51	SPTB	c.759delC/p.E254Kfs*27	(1)
52	SPTB	c.763+1G>A	(1)
53	SPTB	c.877_887delinsAGAC/p.V293Rfs*9	(1)
54	SPTB	c.1920G>A/p.W640*	(1)
55	SPTB	c.1955G>A/p.W652*	(1)
56	SPTB	c.3111dupA/p.H1038Tfs*35	(1)
57	SPTB	c.3916C>T/p. R1306*	(1)
58	SPTB	c.4267C>T/p. R1423*	(1)
59	SPTB	c.4735C>T/p.R1579*	(1)
60	SPTB	c.4973+5G>A	(1)
61	SPTB	c.6238C>T/p.Q2080*	(1)
62	SPTB	c.6747C>G/p.Y2249*	(1)
63	SLC4A1	c.1388G>A/p.G463D	(1)
64	SLC4A1	c.1468C>T/p.R490C	(1)
65	SLC4A1	c.1800G>T/p.K600N	(1)
66	ANK1	c.2T>G/p.M1R	(11)
67	ANK1	c.28-2A>G	(11)
68	ANK1	c.191T>C/p.L64P	(11)
69	ANK1	c.841C>T/p.R281*	(11)
70	ANK1	c.923dupC/p.I309Nfs*47	(11)
71	ANK1	c.1305+5G>A	(11)

72	ANK1	c.1420delC/p.L474Ffs*13	(11)
73	ANK1	c.1616dupC/p.L540Sfs*81	(11)
74	ANK1	c.2394_2397 del/p.S799Ifs*5	(11)
75	ANK1	c.2559-2A>G	(11)
76	ANK1	c.2891dupC/p.L965Tfs*152	(11)
77	ANK1	c.3179C>T/p.P1060L	(11)
78	ANK1	c.4000C>T/p.R1334*	(11)
79	ANK1	c.4387_4390delAACA/p.N1463Wfs*17	(11)
80	ANK1	c.4462C>T/p.R1488*	(11)
81	ANK1	c.4462C>T/p.R1488*	(11)
82	SPTB	c.647G>A/p.R216Q	(11)
83	SPTB	c.1412C>T/p.T471M	(11)
84	SPTB	c.1816C>T/p.Q606*	(11)
85	SPTB	c.1912C>T/p.R638*	(11)
86	SPTB	c.2533C>T/p.Q845*	(11)
87	SPTB	c.4105A>G/p.K1369E	(11)
88	SPTB	c.4267C>T/p. R1423*	(11)
89	SPTB	c.4403_4404 insGA/p.R1469Efs*20	(11)
90	SPTB	c.4538_4539 delTG/p.V1513Afs*13	(11)
91	SPTB	c.4735C>T/p.R1579*	(11)
92	SPTB	c.4804G>T/p.E1602*	(11)
93	SPTB	c.5038delG/p.E1680Kfs*32	(11)
94	SPTB	c.5212G>T/p.E1738*	(11)
95	SPTB	c.5551C>T/p.Q1851*	(11)
96	SPTB	c.5937+1G>C	(11)
97	SPTB	c.5938-2A>T	(11)
98	SLC4A1	c.332G>A/p.R111H	(11)
99	SLC4A1	c.448C>T/p.R150*	(11)
100	SLC4A1	c.1363G>A/p.P455R	(11)
101	ANK1	c.709C>T/p.Q237*	(2)
102	ANK1	c.856C> T/p.R286*	(2)
103	ANK1	c.2950C>T/p.Q984*	(2)
104	ANK1	c.3754C>T/p.R1252*	(2)
105	SPTB	c.3984G>A/p.W1328*	(2)
106	SPTB	c.4978C>T/p.Q1660*	(2)
107	SPTB	c.5933_5934delAG/p.E1978Gfs*18	(2)
108	ANK1	c.858_862delAATCT/p.I287Rfs*67	(12)

109	ANK1	c.3275delA/p.Q1092Rfs*13	(12)
110	ANK1	c.4306C>T/p.R1436*	(12)
111	SPTB	c.318delG/p.K107Rfs*33	(12)
112	SPTB	c.3484delC/p.H1162Tfs*64	(12)
113	ANK1	c.824C>G/p.P275R	(13)
114	SPTB	c.211G>A/p.V71M	(13)
115	SPTB	c.4181G>A/p.W1394*	(13)
116	SPTB	c.4973+5G>A	(13)
117	SLC4A1	c.256-258delinsTTCTC/p.E86Ffs*24	(13)
118	ANK1	c.830A>G/p.H277R	(14)
119	ANK1	c.985G>C/p.A329P	(14)
120	SLC4A1	c.37G>A/p.Q13K	(14)
121	SLC4A1	c.340T>C/p.F114L	(14)
122	ANK1	c.841C>T/p.R281* c.5572C>G/p.L1858V	(3)
123	SPTA1	c.161A>C/p.H54P 6531-12C>T	(15)
124	SLC4A1	c.113A>C/p.D38A	(16)
125	SLC4A1	c.166A>G/p.K56E	(16)
126	SPTB	c.C4873T/p.R1625*	(17)
127	SLC4A1	c.1469G>A/p.R1625*	(17)
128	SPTB	c.5650G>C/p.A1884P	(18)
129	ANK1	c.796G>T/p.E266*	(19)
130	ANK1	c.5422delG/p.E1808Sfs*3	(20)
131	SPTB	c.2413C>T/p.Q805*	(21)
132	SPTB	c.764-1G>C	(22)
133	ANK1	c.1801-1C>G	(23)
134	SPTB	c.6095T>C/p.L2032P	(24)
135	ANK1	c.229-2A>C	(25)
136	ANK1	c.4276C>T/p.R1426*	(26)

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