



CORRECTION



Correction to: The value of single-molecule real-time technology in the diagnosis of rare thalassemia variants and analysis of phenotype–genotype correlation

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In the original Table 1 of this article, the names of seventh and eighth columns were misplaced to each other. The name of

seventh column should be “Detection range of conventional technology”, while the name of eighth column should be “Detection range of SMRT”. The corrected table is shown below. The conclusions of the paper are not affected.

Table 1. Thalassemia variants identified by SMRT and conventional technology

| Common name | HGVS name | Clinical significance | Allele frequency ^a | Occurrence ^a | n (%) | Detection range of conventional technology | Detection range of SMRT | Verification method | Reference |
|--|--|------------------------|-------------------------------|---|-------------|--|-------------------------|---------------------|------------|
| - α ² | N/A1 | Pathogenic | None | Chinese East Asian Indian | 130 (26.80) | YES | YES | Gap-PCR/MLPA | [2] |
| - α 3.7 | NG_000006.1:g.34164_37967del3804 | Pathogenic | None | African, Far East, Indian, Mediterranean | 95 (19.59) | YES | YES | Gap-PCR/MLPA | [2] |
| --SEA | NG_000006.1:g.26264_45564del19301 | Pathogenic | None | East Asian | 94 (19.38) | YES | YES | Gap-PCR/MLPA | [2] |
| --THAI | NC_000016.10:g.149863_183312del | Pathogenic | None | Thai | 2 (0.41) | YES | YES | Gap-PCR/MLPA | [2] |
| - α ² -4 | N/A1 | Pathogenic | None | Chinese | 1 (0.21) | NO | YES | Gap-PCR/MLPA | [8] |
| HS-40 deletion | NC_000016.10:g.(47217_113592)_(113687_143639)del | Pathogenic | None | Chinese | 2 (0.41) | YES | NO | MLPA | [9] |
| HBG1-HBG2 | N/A1 | Uncertain-Significance | None | Chinese | 1 (0.21) | YES | NO | MLPA | This study |
| Hk $\alpha\alpha$ | N/A1 | Uncertain-Significance | None | Chinese | 6 (1.24) | NO | YES | Gap-PCR | [12] |
| $\alpha\alpha\alpha$ ^{anti-4.2} | N/A1 | Uncertain-Significance | None | Chinese | 2 (0.41) | NO | YES | Gap-PCR | [10, 11] |
| $\alpha\alpha\alpha$ ^{anti-3.7} | N/A1 | Uncertain-Significance | None | Chinese | 2 (0.41) | NO | YES | Gap-PCR | [10, 11] |
| CD142(TAA>CAA) | HBA2:c.427T>C | Pathogenic | 14/248854, GnomAD_exome | Arabian Cambodian Chinese Greek Indian Indonesian Laotian Malaysian Sicilian Vietnamese | 18 (3.71) | YES | YES | RDB/Sanger | [2] |
| CD125(CTG>CCG) | HBA2:c.377T>C | Pathogenic | 1/136864, GnomAD | Chinese | 3 (0.62) | YES | YES | RDB/Sanger | [2] |
| CD122(CAC>CAG) | HBA2:c.369C>G | Uncertain-Significance | 19/116218, ExAC | Chinese Laotian | 2 (0.41) | YES | YES | RDB/Sanger | [2] |
| CD11(AAG>CAG) | HBA1:c.34A>C | Uncertain-Significance | 19/116218, ExAC | Chinese | 1 (0.21) | NO | YES | Sanger | [13] |
| CD16(AAG>AAC) | HBA1:c.51G>C | Uncertain-Significance | None | Chinese Pakistani | 1 (0.21) | NO | YES | Sanger | [14] |
| CD27(AAG>AAT) | HBA1:c.84G>T | Uncertain-Significance | None | Chinese | 2 (0.41) | NO | YES | Sanger | [15] |
| CD6(GAC>TAC) | HBA1:c.19G>T | Uncertain-Significance | None | Vietnamese | 1 (0.21) | NO | YES | Sanger | [16] |

Table 1. continued

| Common name | HGVS name | Clinical significance | Allele frequency ^a | Occurrence ^a | n (%) | Detection range of conventional technology | Detection range of SMRT | Verification method | Reference |
|------------------|----------------------|------------------------------|-------------------------------|--|-----------|--|-------------------------|---------------------|-----------|
| CD18(GGC>CGC) | HBA1:c.55G>C | Uncertain-Significance | 2/138328, GnomAD | Chinese Indian Saudi Arabian | 4 (0.82) | NO | YES | Sanger | [17] |
| Init CD(ATG>A-G) | HBA2:c.2delT | Pathogenic | None | Vietnamese | 1 (0.21) | NO | YES | Sanger | [18] |
| Init CD(ATG>ACG) | HBA2:c.2T>C | Pathogenic | 0/654, ALFA | Italian | 1 (0.21) | NO | YES | Sanger | [19] |
| CD17(GTC>TTC) | HBA2:c.52G>T | Uncertain-Significance | None | Chinese | 1 (0.21) | NO | YES | Sanger | [20] |
| CD30(GAG>CAG) | HBA2:c.91G>C | Uncertain-Significance | 1/264690, TOPMED | Chinese | 3 (0.62) | NO | YES | Sanger | [21] |
| -22 C>T | HBA2:c.-59C>T | Pathogenic-Likely-Pathogenic | None | Nedlands | 1 (0.21) | NO | YES | Sanger | [22] |
| CD85(GAC>AAC) | HBA2:c.256G>A | Uncertain-Significance | None | English | 1 (0.21) | NO | YES | Sanger | [23] |
| CD41/42(-TTCT) | HBB:c.126_129delCTTT | Pathogenic | 7/140174, GnomAD | Chinese 41.84% English 4.35% Indonesian 1.69% Japanese 5.99% Korean 4.17% Malaysian 26.32% Pakistani 6.7% Punjabi 13.22% Singapore 37.59% Taiwanese 30.63% Thai 37.24% | 22 (4.54) | YES | YES | RDB/Sanger | [2] |
| CD17(AAG>TAG) | HBB:c.52A>T | Pathogenic | 3/140268, GnomAD | Chinese 14.1% Indonesian 1.69% Japanese 0.32% Korean 16.67% Malaysian 5.26% Singapore 9.02% Taiwanese 8.13% Thai 18.56% | 15 (3.09) | YES | YES | RDB/Sanger | [2] |
| -28(A>G) | HBB:c.-78A>G | Pathogenic-Likely-Pathogenic | 1/140226, GnomAD | Chinese 12.31% Japanese 0.32% Malaysian 6.43% Taiwanese 9.38% Thai 6.83% | 10 (2.06) | YES | YES | RDB/Sanger | [2] |
| CD26(GAG>AAG) | HBB:c.79G>A | Pathogenic | 9/140272, GnomAD | Thai 0.12% | 5 (1.03) | YES | YES | RDB/Sanger | [2] |
| CD71/72(+A) | HBB:c.216_217insA | Pathogenic | 2/251430, GnomAD_exome | Chinese East Asian | 3 (0.62) | YES | YES | RDB/Sanger | [2] |

Table 1. continued

| Common name | HGVs name | Clinical significance | Allele frequency ^a | Occurrence ^a | n (%) | Detection range of conventional technology | Detection range of SMRT | Verification method | Reference |
|-----------------|------------------|------------------------|-------------------------------|---|-----------|--|-------------------------|---------------------|-----------|
| IVS-II-654(C>T) | HBB:c.316-197C>T | Pathogenic | 7/140170, GnomAD | Chinese 21.37% Indonesian 11.86% Japanese 11.99% Malaysian 10.53% Russian 1.52% Singapore 25.56% Taiwanese 46.25% Thai 5.13% | 2 (0.41) | YES | YES | RDB/Sanger | [2] |
| CD14/15(+G) | HBB:c.45dupG | Pathogenic | 1/251224, GnomAD_exome | Chinese Thai 0.12% | 1 (0.21) | YES | YES | RDB/Sanger | [2] |
| -29(A>G) | HBB:c.-79A>G | Pathogenic | 127/140260, GnomAD | Algerian 3.8% Black 59.38% Chinese 2.37% Malaysian 0.58% Taiwanese 0.63% | 1 (0.21) | YES | YES | RDB/Sanger | [2] |
| CD27/28(+C) | HBB:c.84_85insC | Pathogenic | 1/264690, TOPMED | Chinese 0.59% Singapore 0.75% Taiwanese 2.5% Thai 0.24% | 1 (0.21) | YES | YES | RDB/Sanger | [2] |
| IVS-I-5(G>C) | HBB:c.92+5G>C | Pathogenic | 1/140258, GnomAD | Frequent in Asian Indian, UAE, and East Asian populations | 1 (0.21) | YES | YES | RDB/Sanger | [2] |
| -50(G>A) | HBB:c.-100G>A | Uncertain-Significance | 2/140260, GnomAD | Chinese | 1 (0.21) | NO | YES | Sanger | [24] |
| -86 (C>G) | HBB:c.-136C>G | Pathogenic | 0/78698, GnomAD | Lebanese, Thai 0.24% | 1 (0.21) | NO | YES | Sanger | [25] |
| IVS-II-5(G>C) | HBB:c.315+5G>C | Pathogenic | 1/140204, GnomAD | Chinese 0.15% | 10 (2.06) | NO | YES | Sanger | [26] |
| CD126 (GTG>GGG) | HBB:c.380T>G | Pathogenic | 1/140228, GnomAD | German Italian Thai | 1 (0.21) | NO | YES | Sanger | [27] |
| -31(A>C) | HBB:c.-81A>C | Pathogenic | 1/264690, TOPMED | Italian Chinese | 1 (0.21) | NO | YES | Sanger | [22] |
| CD30 (A>G) | HBB:c.91A>G | Likely-Pathogenic | None | Sephardic Jewish | 5 (1.03) | NO | YES | Sanger | [28] |
| CD56 (GGC>GAC) | HBB:c.170G>A | Likely-Benign | 5/251448, GnomAD_exome | Found in Thai, Indonesian, Black, and Chinese families | 5 (1.03) | NO | YES | Sanger | [29] |
| CD 64 (GGC>AGC) | HBB:c.193G>A | Likely-Benign | None | Chinese | 1 (0.21) | NO | YES | Sanger | [30] |

Table 1. continued

| Common name | HGVs name | Clinical significance | Allele frequency ^a | Occurrence ^a | n (%) | Detection range of conventional technology | Detection range of SMRT | Verification method | Reference |
|-------------------|-------------------|------------------------|-------------------------------|---------------------------------------|-----------|--|-------------------------|---------------------|------------|
| CD77 (CAC>TAC) | HBB:c.232C>T | Likely-Benign | 1/251428, GnomAD_exom | Caucasian Indonesian Japanese Swedish | 1 (0.21) | NO | YES | Sanger | [31] |
| -198A>G | HBB:c.-248A>G | Uncertain-Significance | None | This study | 3 (0.62) | NO | YES | Sanger | This study |
| CD113(GTG>GAG) | HBB:c.341T>A | Pathogenic | 2/140270, GnomAD | American Chinese | 14 (2.89) | NO | YES | Sanger | [32] |
| CD143(CAC>CGC) | HBB:c.431A>G | Likely-Pathogenic | 1/251362, GnomAD_exome | American Italian | 1 (0.21) | NO | YES | Sanger | [33] |
| IVS- II-806 (G>C) | HBB:c.316-45G>C | Benign-Likely-Benign | 34/140174, GnomAD | Chinese | 3 (0.62) | NO | YES | Sanger | This study |
| IVS- II-672 (A>C) | HBB:c.316-179A>C | Benign-Likely-Benign | None | Chinese | 1 (0.21) | NO | YES | Sanger | This study |
| IVS -II-308 (-A) | HBB:c.315+308delA | Benign-Likely-Benign | None | Chinese | 1 (0.21) | NO | YES | Sanger | This study |
| Total | | | | | 485 (100) | | | | |

Gap-PCR Gap- polymerase chain reaction, MLPA multiple ligation probe amplification technology, RDB reverse dot blot.

^aData come from HbVar (<https://globin.bx.psu.edu/hbvar/hbvar.html>), genomAD (<https://gnomad.broadinstitute.org/>) and dbSNP (<https://www.ncbi.nlm.nih.gov/snp/>).