**S1 Table:** **The genotypes, phenotypes, IC strip test results and hematologic parameters (MCV and MCH) of the 195 blood samples.**

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| **Samples** | **Genotype** | **Phenotype\*** | **IC stripe** | | **MCV/MCH\*\*** | |
| **ζ-globin** | **Hb Bart's** | **High** | **Low** |
| 1 | --SEA/αCSα | HbH-CS disease | Positive | Positive |  | 🗸 |
| 2 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 3 | --SEA/αCSα | HbH-CS disease | Positive | Positive |  | 🗸 |
| 4 | --SEA/αCSα | HbH-CS disease | Positive | Positive |  | 🗸 |
| 5 | --SEA/αCSα | HbH-CS disease | Positive | Positive |  | 🗸 |
| 6 | --SEA/-α3.7 | HbH disease | Positive | Positive |  | 🗸 |
| 7 | --THAI/-α3.7 | HbH disease | Negative | Positive |  | 🗸 |
| 8 | --THAI/-α3.7 | HbH disease | Negative | Positive |  | 🗸 |
| 9 | --SEA/-α3.7 | HbH disease | Positive | Positive |  | 🗸 |
| 10 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 11 | β17/βN, -α3.7/αα | Beta-thalassemia heterozygote with alpha-thalassemia 2 trait | Positive | Positive |  | 🗸 |
| 12 | β17/βN, αα/αα | Beta-thalassemia heterozygote | Negative | Negative |  | 🗸 |
| 13 | --SEA/αCSα | HbH-CS disease | Positive | Positive |  | 🗸 |
| 14 | βE/βN, --SEA/αCSα | CSEABart's disease | Positive | Positive |  | 🗸 |
| 15 | βE/βE, -α3.7/αα | Homozygous HbE with alpha-thalassemia 2 trait | Positive | Positive |  | 🗸 |
| 16 | β17/βN, αα/αα | Beta-thalassemia heterozygote | Negative | Negative |  | 🗸 |
| 17 | --THAI/-α3.7 | HbH disease | Negative | Positive |  | 🗸 |
| 18 | βE/βN, αα/αα | HbE heterozygote | Negative | Negative | 🗸 |  |
| 19 | αCSα/αα | Hb Constant Spring heterozygote | Negative | Positive | 🗸 |  |
| 20 | βE/βN, αα/αα | HbE heterozygote | Negative | Negative | 🗸 |  |
| 21 | β4bp/βN, αα/αα | Beta-thalassemia heterozygote | Positive | Negative |  | 🗸 |
| 22 | β4bp/βE, αα/αα | Beta-thalassemia/HbE disease | Negative | Negative |  | 🗸 |
| 23 | --SEA/αCSα | HbH-CS disease | Positive | Positive |  | 🗸 |
| 24 | αCSα/αα | Hb Constant Spring heterozygote | Negative | Positive | 🗸 |  |
| 25 | β4bp/β4bp, αα74/αα | Beta-thalassemia homozygote with alpha-thalassemia 2 trait | Negative | Positive |  | 🗸 |
| 26 | --SEA/αCSα | HbH-CS disease | Positive | Positive |  | 🗸 |
| 27 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 28 | -α3.7/αα | Alpha-thalassemia 2 heterozygote | Negative | Positive | 🗸 |  |
| 29 | βE/βE, -α3.7/αα | Homozygous HbE with alpha-thalassemia 2 trait | Positive | Negative |  | 🗸 |
| 30 | --SEA/-α3.7 | HbH disease | Positive | Positive |  | 🗸 |
| 31 | --SEA/αCSα | HbH-CS disease | Positive | Positive |  | 🗸 |
| 32 | --THAI/αCSα | HbH-CS disease | Negative | Positive |  | 🗸 |
| 33 | --SEA/αCSα | HbH-CS disease | Positive | Positive |  | 🗸 |
| 34 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 35 | -α3.7/αα | Alpha-thalassemia 2 heterozygote | Positive | Negative | 🗸 |  |
| 36 | --SEA/-α3.7 | HbH disease | Positive | Positive |  | 🗸 |
| 37 | --SEA/-α4.2 | HbH disease | Positive | Positive |  | 🗸 |
| 38 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 39 | --SEA/-α3.7 | HbH disease | Positive | Positive |  | 🗸 |
| 40 | --SEA/αCSα | HbH-CS disease | Positive | Positive |  | 🗸 |
| 41 | --SEA/αCSα | HbH-CS disease | Positive | Positive |  | 🗸 |
| 42 | --SEA/αCSα | HbH-CS disease | Positive | Positive |  | 🗸 |
| 43 | βE/βN, --SEA/αα | HbE heterozygote with alpha-thalassemia 1 trait | Positive | Positive |  | 🗸 |
| 44 | βE/βN, --SEA/αCSα | CSEABart's disease | Positive | Positive |  | 🗸 |
| 45 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 46 | βIVSI-1/βE, --SEA/αα | Beta-thalassemia/HbE disease with alpha-thalassemia 1 trait | Positive | Negative |  | 🗸 |
| 47 | -α3.7/αα | Alpha-thalassemia 2 heterozygote | Positive | Negative | 🗸 |  |
| 48 | --SEA/-α3.7 | HbH disease | Positive | Positive |  | 🗸 |
| 49 | --SEA/-α3.7 | HbH disease | Positive | Positive |  | 🗸 |
| 50 | β4bp/βE, --SEA/αα | Beta-thalassemia/HbE disease with alpha-thalassemia 1 trait | Positive | Positive |  | 🗸 |
| 51 | --SEA/-α3.7 | HbH disease | Positive | Positive |  | 🗸 |
| 52 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 53 | --SEA/αCSα | HbH-CS disease | Positive | Positive |  | 🗸 |
| 54 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 55 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 56 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 57 | βE/βN, --SEA/αα | HbE heterozygote with alpha-thalassemia 1 trait | Positive | Positive |  | 🗸 |
| 58 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 59 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 60 | --SEA/-α3.7 | HbH disease | Positive | Positive |  | 🗸 |
| 61 | --SEA/-α4.2 | HbH disease | Positive | Positive |  | 🗸 |
| 62 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 63 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 64 | βE/βE, αα/αα | Homozygous HbE | Negative | Negative |  | 🗸 |
| 65 | βE/βN, αα/αα | HbE heterozygote | Negative | Negative | 🗸 |  |
| 66 | -α3.7/αα | Alpha-thalassemia 2 heterozygote | Negative | Negative | 🗸 |  |
| 67 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 68 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 69 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 70 | βE/βN, αα/αα | HbE heterozygote | Negative | Negative | 🗸 |  |
| 71 | βE/βN, αCSα/αα | HbE heterozygote with Hb Constant Spring heterozygote | Negative | Positive | 🗸 |  |
| 72 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 73 | -α3.7/αα | Alpha-thalassemia 2 heterozygote | Positive | Positive | 🗸 |  |
| 74 | βE/βE, αα/αα | Homozygous HbE | Negative | Positive |  | 🗸 |
| 75 | β19/βN, αα/αα | Beta-thalassemia heterozygote | Negative | Negative |  | 🗸 |
| 76 | βE/βE, αα/αα | Homozygous HbE | Negative | Negative |  | 🗸 |
| 77 | --SEA/αCSα | HbH-CS disease | Positive | Positive |  | 🗸 |
| 78 | --SEA/αCSα | HbH-CS disease | Positive | Positive |  | 🗸 |
| 79 | αCSα/αα | Hb Constant Spring heterozygote | Negative | Positive | 🗸 |  |
| 80 | βE/βN, --SEA/αCSα | CSEABart's disease | Positive | Positive |  | 🗸 |
| 81 | αCSα/αα | Hb Constant Spring heterozygote | Negative | Positive | 🗸 |  |
| 82 | βE/βN, --SEA/αα | HbE heterozygote with alpha-thalassemia 1 trait | Positive | Positive |  | 🗸 |
| 83 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 84 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 85 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 86 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 87 | βE/βN, -α3.7/αα | HbE heterozygote with alpha-thalassemia 2 heterozygote | Negative | Negative | 🗸 |  |
| 88 | -α3.7/αα | Alpha-thalassemia 2 heterozygote | Positive | Negative | 🗸 |  |
| 89 | -α3.7/αα | Alpha-thalassemia 2 heterozygote | Negative | Positive | 🗸 |  |
| 90 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 91 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 92 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 93 | -α3.7/αα | Alpha-thalassemia 2 heterozygote | Negative | Negative | 🗸 |  |
| 94 | βE/βN, --SEA/αCSα | CSEABart's disease | Positive | Positive |  | 🗸 |
| 95 | βE/βN, --SEA/αα | HbE heterozygote with alpha-thalassemia 1 trait | Positive | Negative |  | 🗸 |
| 96 | αInt (-T)α/αα | Alpha-thalassemia 2 heterozygote | Positive | Positive | 🗸 |  |
| 97 | --SEA/αInt (-T)α/αα | HbH disease | Positive | Positive |  | 🗸 |
| 98 | -α3.7/-α3.7 | Alpha-thalassemia 2 homozygote | Positive | Positive |  | 🗸 |
| 99 | -α3.7/αα | Alpha-thalassemia 2 heterozygote | Negative | Negative | 🗸 |  |
| 103 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 101 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 102 | βE/βN, αα/αα | HbE heterozygote | Negative | Negative | 🗸 |  |
| 103 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 104 | αCSα/αα | Hb Constant Spring heterozygote | Negative | Positive | 🗸 |  |
| 105 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 106 | βE/βN, --SEA/αα | HbE heterozygote with alpha-thalassemia 1 trait | Positive | Positive |  | 🗸 |
| 107 | --SEA/-α3.7 | HbH disease | Positive | Positive |  | 🗸 |
| 108 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 109 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 110 | βE/βN, -α3.7/-α3.7 | HbE heterozygote with alpha-thalassemia 2 homozygote | Negative | Positive |  | 🗸 |
| 111 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 112 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 113 | αα/αα | Normal Hemoglobin type | Negative | Positive | 🗸 |  |
| 114 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 115 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 116 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 117 | αα/αα | Normal Hemoglobin type | Positive | Negative | 🗸 |  |
| 118 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 119 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 120 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 121 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 122 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 123 | αα/αα | Normal Hemoglobin type | Positive | Negative | 🗸 |  |
| 124 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 125 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 126 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 127 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 128 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 129 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 130 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 131 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 132 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 133 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 134 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 135 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 136 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 137 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 138 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 139 | αα/αα | Normal Hemoglobin type | Positive | Negative | 🗸 |  |
| 140 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 141 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 142 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 143 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 144 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 145 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 146 | -α3.7/-α4.2 | Alpha-thalassemia 2 homozygote | Negative | Positive |  | 🗸 |
| 147 | -α3.7/αα | Alpha-thalassemia 2 heterozygote | Negative | Negative | 🗸 |  |
| 148 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 149 | βE/βN, --SEA/-α3.7 | EABart's disease | Positive | Positive |  | 🗸 |
| 150 | βE/βN, --SEA/-αT | EABart's disease | Positive | Negative |  | 🗸 |
| 151 | αα/αα | Normal Hemoglobin type | Positive | Negative | 🗸 |  |
| 152 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 153 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 154 | αα/αα | Normal Hemoglobin type | Positive | Negative | 🗸 |  |
| 155 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 156 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 157 | -α3.7/-α3.7 | Alpha-thalassemia 2 homozygote | Negative | Positive |  | 🗸 |
| 158 | -α3.7/αα | Alpha-thalassemia 2 heterozygote | Positive | Positive | 🗸 |  |
| 159 | αCSα/αα | Hb Constant Spring heterozygote | Negative | Positive | 🗸 |  |
| 160 | αCSα/αα | Hb Constant Spring heterozygote | Positive | Positive | 🗸 |  |
| 161 | βE/βN, -α3.7/αα | HbE heterozygote with alpha-thalassemia 2 heterozygote | Positive | Negative | 🗸 |  |
| 162 | βE/βN, --SEA/αCSα | CSEABart's disease | Positive | Positive |  | 🗸 |
| 163 | βE/βE, αα/αα | Homozygous HbE | Positive | Negative |  | 🗸 |
| 164 | βE/βN, αα/αα | HbE heterozygote | Positive | Negative | 🗸 |  |
| 165 | βE/βN, αα/αα | HbE heterozygote | Negative | Positive | 🗸 |  |
| 166 | αCSα/αCSα | Homozygous Constant Spring | Negative | Positive | 🗸 |  |
| 167 | αCSα/αα | Hb Constant Spring heterozygote | Negative | Positive | 🗸 |  |
| 168 | βE/βN, αα/αα | HbE heterozygote | Negative | Negative | 🗸 |  |
| 169 | β4bp/βN, -α3.7/αα | Beta-thalassemia heterozygote with alpha-thalassemia 2 trait | Negative | Negative |  | 🗸 |
| 170 | -α3.7/-α3.7 | Alpha-thalassemia 2 homozygote | Positive | Positive |  | 🗸 |
| 171 | -α3.7/αα | Alpha-thalassemia 2 heterozygote | Negative | Positive | 🗸 |  |
| 172 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 173 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 174 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 175 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 176 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 177 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 178 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 179 | βE/βN, -α3.7/-α3.7 | HbE heterozygote with alpha-thalassemia 2 homozygote | Negative | Positive |  | 🗸 |
| 180 | -α3.7/αα | Alpha-thalassemia 2 heterozygote | Negative | Negative | 🗸 |  |
| 181 | -α3.7/-α3.7 | Alpha-thalassemia 2 homozygote | Positive | Positive |  | 🗸 |
| 182 | -α3.7/αCSα | Alpha-thalassemia 2 heterozygote with Hb CS heterozygote | Positive | Positive | 🗸 |  |
| 183 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 184 | -α3.7/-α4.2 | Alpha-thalassemia 2 homozygote | Negative | Positive |  | 🗸 |
| 185 | -α3.7/αα | Alpha-thalassemia 2 heterozygote | Positive | Positive | 🗸 |  |
| 186 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 187 | αα/αα | Normal Hemoglobin type | Negative | Negative | 🗸 |  |
| 188 | -α4.2/αα | Alpha-thalassemia 2 heterozygote | Negative | Negative | 🗸 |  |
| 189 | βE/βN, αα/αα | HbE heterozygote | Positive | Negative | 🗸 |  |
| 190 | βE/βN, -α3.7/αα | HbE heterozygote with alpha-thalassemia 2 heterozygote | Negative | Negative | 🗸 |  |
| 191 | -α3.7/αα | Alpha-thalassemia 2 heterozygote | Negative | Positive | 🗸 |  |
| 192 | αCSα/αα | Hb Constant Spring heterozygote | Negative | Positive | 🗸 |  |
| 193 | --SEA/αα | Alpha-thalassemia 1 heterozygote | Positive | Positive |  | 🗸 |
| 194 | βE/βN, αα/αα | HbE heterozygote | Negative | Positive | 🗸 |  |
| 195 | βE/βE, αα/αα | Homozygous HbE | Negative | Negative |  | 🗸 |

\* Alpha-thalassemia 1 heterozygote (α0-thalassemia heterozygote); Alpha-thalassemia 2 heterozygote (α+-thalassemia heterozygote)

\*\* High = MCV ≥ 80 fL and MCH ≥ 27 pg (High MCV/MCH); Low = MCV < 80 fL or MCH < 27 pg (Low MCV/MCH)

One hundred ninety-five subjects with various thalassemia and normal hemoglobin were recruited in this study. The genotypes and phenotypes of each sample are shown. Samples with MCV ≥ 80 fL and MCH ≥ 27 pg (High MCV/MCH) or MCV < 80 fL or MCH < 27 pg (Low MCV/MCH) are indicated. The IC strip test results (+, positive; -, negative) of each subject are also indicated.