Thalassemia Carrier Screening
Consent Form

Patient Information (Completed by the patient) **Sample No. _____**

Name		DOB (d/m/y)	/ /
ID/Passport No.		Gender	☐ Male ☐ Female
Mobile Phone No.		Genetic Locus in Family	☐ Known ☐Unknown
Address			

Clinical Information (Recorded by health care providers)

Medical Record No.		Collection Date (d/m/y)	/ /
Hospital/Clinic		Physician (Signature)	
Specimen Type	☐ Blood(EDTA) ☐ Amniotic Fluid: _____ml ☐ Villi ☐ Cord Blood ☐ Placenta ☐ Cord ☐ Other_____		
Pedigree and Clinical Details			

I, the undersigned, understand the Thalassemia Carrier Screening serve as an identification of common thalassemia mutations. I hereby fully understand, agree and undertake the following:

1. This test identifies several indicators of red blood cell and serum in peripheral blood, and also domestically common genotypes of thalassemia.
2. There are several different and distinct alpha-thalassemia subtypes. This test detects the 7 most common deletions of HBA1 and HBA2 genes (--SEA, --THAI, --FIL, --MED, -α20.5, -α3.7, -α4.2) and detects 2 common point mutations in HBA2 gene (Hb Constant Spring and Hb Quong Sze).
3. More than 95% of beta-thalassemia is caused by a point mutation or small deletion/insertion leading to frameshift in the HBB gene. This test focuses on screening common mutations in the promoter area, coding regions, intron-exon boundaries, especially common mutations in Asian region (-28, initiation codon, codon 15, codon 17, codon 26, codon 27/28, codon 41/42, codon 43, codon 71/72, IVSI-1, IVSI-5, IVSII-654).
4. The test cannot detect large beta globin gene deletion, rare alpha globin gene deletion, rare non-deletion mutations, gene duplications, and mutations of the regulatory region.
5. Iron deficiency anemia is the most common type of anemia, and thalassemia trait can be confused with iron-deficiency anemia because MCV and MCH are lower than normal in both conditions. Therefore, this test may be harder to distinguish thalassemia from iron-deficiency anemia in patients with both types of anemia.
6. Besides, diagnostic errors may occur as a result of gene recombination, cell contamination, and a number of other complex factors.
7. For all the reasons above, the accuracy of this test is approximately 97%.
8. I hereby agree that the hospital/clinic and Sofiva Genomics may collect, process or use my personal information such as medical records, medical treatment, genetic information and health examination records under the specific purpose of medical care, health treatment etc.
9. I ☐agree / ☐do not agree to allow the remainder of my sample to be used for research purposes. (Lack of response indicates consent.)
10. According to my situation, the physician has answered all my questions and adequately explained to me (included but not restricted to the information about the necessary, process, potential risk and successful rate of this test as well as the risk of other screening tests).
11. I fully understand the above terms, statements and declarations, and I agree to have this test performed at my own expense. I understand and accept that this test may be the most appropriate choice at this time, but it cannot guarantee the prevention of the tested disorders.

Signature, Date (dd/mm/yyyy)

Copy 1: Laboratory Retain (White) Copy 2: Hospital/Clinic Retain (Blue) Copy 3: Patient Retain (Red)