Sickle Cell and Other Hemoglobinopathies Mutation Panel

Test Code	D0408
Test Summary	Testing of point mutations for Sickle Cell and Other Hemoglobinopathies
Turn-Around-Time (TAT)*	10 - 12 days
Acceptable Sample Types	Dried Blood Spots
Acceptable Billing Types	Self (patient) Payment Institutional Billing Commercial Insurance

Indications for Testing

Symptoms of Sickle Cell and Other Hemoglobinopathies

Test Description

Testing for Hb S (173A>T), Hb C (172G>A), Hb E (232G>A), Hb D (121G>C) and Hb O (121G>A); ? Thalassaemias:: -29A>G, -88C>T, and IVS1+6T>C

Condition Description

Sickle Cell Anemia and/or other Hemoglobinopathies

Test Methods and Limitations

DNA analysis is performed to determine the copy number of one of the T-cell Receptor Excision Circles (TRECs) by real-time quantitative PCR and Absolute Quantification analysis. Serial dilutions of an external standard with predefined known concentrations are used to create a standard curve. During PCR amplification, the fluorescence values are measured to analyze quantification data. The point at which the fluorescence of a sample rises above the background fluorescence is called the “crossing point (Cp)” of the sample. The crossing points of standards and unknown samples are then used to determine the concentration of target DNA.

Detailed Sample Requirements

Dried Blood Spots