HemeScreen® CTP Panel – Product Overview

This document provides a general product overview of the HemeScreen CTP Assay. Additional information can be found on Precipio's website at www.precipiodx.com, and the associated IFU (Instructions For Use), available upon request.

Technology Overview	HemeScreen® is a proprietary set of RUO (Research Use Only) reagents used to screen the wild type (Negative) from Mutated (Positive) genes in a simplified workflow relative to alternative molecular testing technologies (RT-PCR or NGS).
CTP	The myelodysplastic syndromes (MDS) comprise a group of hematologic malignancies characterized by clonal hematopoiesis, one or more cytopenias (i.e., anemia, neutropenia, and/or thrombocytopenia), and abnormal cellular maturation.

Genes Tested	Coverage
WT1 Exon 8	c.1142C>A; p.S381*, c.1110dup; p.V371Cfs*14
WT1 Exon 10	c.1384C>T; p.R462W, c.1385G>A; p.R462Q, c.1385G>C; p.R462P, c.1390G>A; p.D464N
ASXL1 Exon 12 codon 591	c.1772dup; p.Y591*
ASXL1 Exon 12 codon 635	c.1900_1922del; p.E635Rfs*15, c.1934dup; p.G646Wfs*12
ASXL1 Exon 12 codon 693	c.2077C>T; p.R693*
ASXL1 Exon 12 codon 808	c.2423del; p.P808Lfs*10
ASXL1 Exon 12 codon 1102	c.3306G>T; p.E1102D
RUNX1 Exon 4	c.167T>C; p.L56S, c.319C>T; p.R107C
RUNX1 Exon 5	c.422C>A; p.S141*, c.485G>A; p.R162K, c.496C>T; p.R166*
RUNX1 Exon 6	c.592G>A; p.D198N, c.602G>A; p.R201Q, c.610C>T; p.R204*
RUNX1 Exon 8	c.958C>T; p.R320*
DNMT3A Exon 23	c.2645G>A; p.R882H, c.2644C>T; p.R882C, c.2644C>A; p.R882S, c.2645G>C; p.R882P, c.2644C>G; p.R882G, c.2645G>T; p.R882L
SF3B1 Exon 15	c.1866G>T; p.E622D, c.1866G>C; p.E622D, c.1873C>T; p.R625C, c.1874G>T; p.R625L, c.1984C>G; p.H662D, c.1986C>G; p.H662Q, c.1986C>A; p.H662Q, c.1996A>C; p.K666Q, c.1996A>G; p.K666E, c.1997A>C; p.K666T, c.1997A>G; p.K666R, c.1998G>T; p.K666N, c.1998G>C; p.K666N
SF3B1 Exon 16	c.2098A>G; p.K700E
SF3B1 Exon 17	c.2225G>A; p.G742D

Results	The results from HemeScreen® CTP are qualitative.
Associated WHO/NCCN Guidelines[1]	MDS with low blasts, ring sideroblasts and wild-type *SF3B1* has a less favourable overall survival and leukaemia-free survival { 25957392 }. The favourable outcome associated with *SF3B1* mutation is lost as soon as an excess of blasts is observed { 34036300 } The spectrum of sub-clonal genetic alterations identified in MDS-*SF3B1* and driving disease progression is limited. Additional mutations in epigenetic regulators, including *DNMT3A*, *TET2* or *ASXL1* do not affect the disease outcome. In contrast, mutations in *TP53*, *RUNX1*, *EZH2* mutations { 32347921 ; 34036300 } are associated with a poorer outcome.

	Specificity	Sensitivity	LOD	Storage
Assay Specifications	>99%	98%	2%	-20 °C

SKU	Product Configuration	Assay Contents			
HS-3P-CTP	3 sample pre-plated plate	Primers/MasterMix Mix	Positive controls	NTC	Wild Type

Instrument Required	HRM-enabled RT-PCR (example ThermoFisher Quantstudio 3 or higher)
Contact	For further questions, contact our technical support team at techsupport@precipiodx.com or call 203-787-7888
Disclaimer	*The information in this document represents the company's best understanding of the technical and regulatory landscape; however, it should not serve as any guidance to any laboratory seeking to implement HemeScreen. Laboratory managers and medical directors should seek their own information independently through their CLIA inspector and any other state and federal regulatory body available.*

1 World Health Organization. World Health Organization. Available at: https://tumourclassification.iarc.who.int/chaptercontent/63/23