HemeScreen® AML Panel – Product Overview

This document provides a general product overview of the HemeScreen AML 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).
AML	Acute myeloid leukemia (AML) is a clonal malignant neoplasm of myeloid cell lineage involving the blood and bone marrow, but other tissue can also be occasionally affected. In the era of personalized precision medicine, molecular changes have been used in AML classification, diagnosis, prognosis, risk stratification, and treatment.

Genes Tested	Coverage
KIT Exon 9	c.1504_1509dup; p.A502_Y503dup
KIT Exon 11	c.1669_1674del;p.W557_K558del,c.1669T>C; p.W557R, c.1669_1683del;p.W557_E561del, c.1669T>G; p.W557G, c.1669T>A; p.W557R, c.1676T>G; p.V559G, c.1676T>A; p.V559D, c.1676T>C; p.V559A, c.1727T>C; p.L576P, c.1679T>A; p.V560D
KIT Exon 13	c.1924A>G; p.K642E, c.1961T>C; p.V654A, Full exon coverage
KIT Exon 17	c.2446G>C; p.D816H, c.2446G>T; p.D816Y, c.2446_2447GA>AT; p.D816I, c.2447A>T; p.D816V, c.2458G>T; p.D820Y, c.2459A>G; p.D820G, c.2464A>T; p.N822Y, c.2466T>G; p.N822K, c.2466T>A; p.N822K, c.2474T>C; p.V825A, c.2467T>G; p.Y823D
IDH1 Exon 4	c.299G>A; p.R100Q, c.298C>T; p.R100*, c.313G>C; p.G105R, c.314G>T; p.G105V, c.314G>A; p.G105D, c.394C>T; p.R132C, c.394C>G; p.R132G, c.394C>A; p.R132S, c.395G>A; p.R132H, c.395G>T; p.R132L, c.395G>C; p.R132P
IDH2 Exon 4	c.418C>G;p.R140G, c.418C>T; p.R140W, c.419G>A; p.R140Q, c.419G>T; p.R140L, c.515G>T; p.R172M, c.514A>T; p.R172W, c.515G>A; p.R172K, c.516G>T; p.R172S, c.516G>C; p.R172S
FLT3 Exon 14	Internal Tandem Duplications
FLT3 Exon 15	Internal Tandem Duplications
FLT3 Exon 16	Internal Tandem Duplications
FLT3 Exon 20	Mutations in codons 835 and 836
CEBPA Exon 1	Mutation screening of entire exon
NPM1 Exon 12	c.860_863dup; p.W288Cfs*12

Results	The results from HemeScreen® AML are qualitative.
Associated WHO/NCCN Guidelines[1]	*Per the WHO:* AML with *BCR*::*ABL1* and AML with *CEBPA* mutation are the only disease types with a defined genetic abnormality that require at least 20% blasts for diagnosis. AML defined by mutations include AML with *NPM1* and AML with *CEBPA* mutation. AML with *NPM1* mutation can be diagnosed irrespective of the blast count, albeit again with emphasis on judicious clinicopathologic correlation. This approach aligns with data showing that cases previously classified as MDS or MDS/MPN with *NPM1* progress to AML in a short period of time. Similar data have emerged from patients with clonal haematopoiesis who acquire *NPM1* mutation. The definition of AML with *CEBPA* mutation has changed to include biallelic (biCEBPA) as well as single mutations located in the basic leucine zipper (bZIP) region of the gene (smbZIP-*CEBPA*).

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

SKU	Product Configuration	Assay Contents			
HS-1P-AML	1 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/63/37