

# A Comprehensive Whole Genome Sequencing Test for Myeloma Care

EASY, NON-INVASIVE BLOOD OR BONE MARROW TEST FOR DEEP GENETIC INSIGHTS TO GUIDE PERSONALIZED THERAPY SELECTION.

## Result Summary

**Abnormal** (Ref. interval: Normal)

## Interpretation

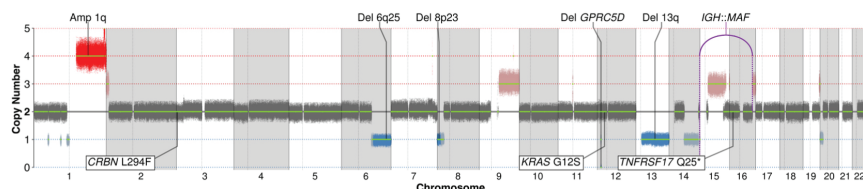
The test detected known genetic biomarkers of multiple myeloma. The alterations IGH::MAF t(14;16) and Amp 1q are characteristic of high-risk multiple myeloma (Refs. 1, 5). Additionally, the following alterations may confer resistance to therapy: Del GPRC5D (hom. del), CRBN p.Leu294Phe and BCMA (biallelic loss via focal del. and p.Gln25fs) (see Result Table). Overall risk should be assessed in context of other risk factors.

Whole genome sequencing was performed on sorted malignant plasma cells, enabling detection of 54 clinically relevant somatic events in multiple myeloma, including translocations (structural variants; SVs), copy number variants (CNVs), single nucleotide variants (SNVs) and small insertions and deletions (indels).

## Specimen Information

Sorted cell count: 250,000  
Tumor cell fraction: 3.8561%  
Estimated purity: 93%  
Median tumor coverage: 67.2x

## Genetic Alterations



GenoPredicta from CTCs enables detection of deletions or mutations of immunotherapy targets. In a clinical sample from a patient with prior BCMA- and GPRC5D-targeting therapies, alterations in both targets were observed by WGS: biallelic deletion of GPRC5D, and deletion and loss-of-function mutation in BCMA.

## Result Table

	Genetic alteration	Therapy association	Refs.
Detected	IGH::MAF t(14;16)	High-risk MM based on IMS-IMWG guidelines	1, 3
	Amp 1q	High-risk MM based on IMS-IMWG guidelines	1
	Del 6q25	Standard-risk MM	31
	Del 13q	Standard-risk MM	21, 30
	Del GPRC5D	Mutations may confer resistance to GPRC5D-targeting therapies	12-15
	Del 8p23	Standard-risk MM	31
	BCMA p.Gln25fs	Mutations may confer resistance to BCMA-targeting therapies	12
	Del BCMA	Mutations may confer resistance to BCMA-targeting therapies	12
	CRBN p.Leu294Phe	Mutations may confer resistance to CRBN-targeting therapies	17
	KRAS p.Gly12Ser	Eligible for RAS-targeting agents	26, 27
Not detected	IGH::FGFR3/NSD2 t(4;14)	High-risk MM based on IMS-IMWG guidelines	1, 2
	IGH::MAFB t(14;20)	High-risk MM based on IMS-IMWG guidelines	1, 4
	IGH::CCND1 t(11;14)	Standard-risk MM, eligible for BCL2 inhibitor	5-7
	Del 1p32	High-risk MM based on IMS-IMWG guidelines	1
	Del 17p	High-risk MM based on IMS-IMWG guidelines	1
	Hyperdiploidy	Standard-risk MM	20, 21
	TP53	High-risk MM based on IMS-IMWG guidelines	1, 8, 9
	CD38	Mutations may confer resistance to CD38-targeting therapies	10, 11
	FCRL5	Mutations may confer resistance to FCRL5-targeting therapies	16
	IKZF1	Resistance to immunomodulatory drugs (e.g., thalidomide, lenalidomide, pomalidomide)	18, 19
Additional negative events tested: IGH/IGK/IGL translocations (excluding partners listed above); CCND1, CCND2, CCND3, FGFR3/NSD2, MAFA, MAFB, MYC; arm-level deletions and/or gains: Del 6q, Del 8p, Gain 11q, Del 16q; markers of hyperdiploidy, e.g., trisomies 3, 5, 7, 9.			
Reference range: "Not detected" for all listed genetic alterations.			

## Discover GenoPredicta's Innovative Myeloma Diagnostics

### 100% CONCORDANCE WITH TRADITIONAL TESTS

Reliable detection of critical genetic abnormalities, including mutations and resistance markers (BCMA, GPRC5D).

### COMPREHENSIVE GENOMIC INSIGHTS

Gain a thorough understanding of your patient's genetic profile for tailored therapies.

### INFORMED PERSONALIZED TREATMENT

Accelerate decision-making, enhance monitoring, and support precision in patient management for improved outcomes.

### MINIMALLY INVASIVE TESTING

Obtain results through blood or bone marrow samples.





## WHAT IS THE DIFFERENCE BETWEEN GENOPREDICTA FOR BLOOD OR BONE MARROW AND HOW DOES IT COMPARE TO FISH?

	FISH	MARROW	BLOOD
Fresh or frozen samples	✓	✓	✓
Minimally invasive	✗	✗	✓
Cell requirements	50/probe	~50	~50
All genetic alterations* from WGS	✗	✗	✓
Satisfies new IMS high-risk criteria (TP53 sequencing)	✗	✓	✓
Detects mutations/deletions of therapeutic targets	✗	✓	✓
Discovers new resistance markers	✗	✓	✓
Representative of all lesions/clones, not just sampled site	✗	✗	✓
Monitoring of treatment response (clonal evolution)	✗	✗	✓
Other blood cancers	✗	✗	✓

GenoPredicta is a cutting-edge test that can replace painful bone marrow biopsies and outdated FISH. The table shows a head-to-head comparison of GenoPredicta with FISH. In the future, GenoPredicta will also be available for other hematological malignancies.

## CAN GENOPREDICTA REPLACE A BONE MARROW BIOPSY?

While the initial diagnosis of multiple myeloma still requires a bone marrow biopsy, future follow-up monitoring may not. Current tests like FISH require painful biopsies to collect malignant plasma cells, making ongoing monitoring difficult. GenoPredicta addresses this issue by collecting as few as ~50 circulating tumor cells from peripheral blood, allowing for better prognostication without the need for a invasive biopsies.

