

Joshua Richter, MD

Clinical Assistant Professor
Rutgers University
New Brunswick, New Jersey
John Theurer Cancer Center
Division of Myeloma
Hackensack University Medical Center
Hackensack, New Jersey

What are the different technologies used to assess cytogenetic risk status?

1. Three tools to assess cytogenetic risk status in multiple myeloma
 - a. Routine karyotyping
 - b. Fluorescence in situ hybridization (FISH)
 - c. Gene expression profiling
2. Routine karyotyping
 - a. 20 cells evaluated during metaphase for all forms of genetic abnormalities
 - b. Abnormalities include additions, deletions, and other genetic problems, including those known in myeloma and those that have no clear relevance in myeloma
 - c. Most coarse approach because it only evaluates 20 cells
3. Fluorescence in situ hybridization (FISH)
 - a. Hundreds of cells evaluated to identify certain abnormalities that have known clinical significance in myeloma
 - b. Panels differ between institutions, but commonly include the known variables:
 - i. del17p
 - ii. 1q addition
 - iii. t(4;14)
 - iv. t(11;14)
4. Gene expression profiling
 - a. Large arrays that evaluate overall gene signatures as to which genes are expressed or not expressed; may have ramifications for patients' ability to stay in remission or have early relapse
 - b. Two main platforms
 - i. Signal Genetics MyPRS® score; a 70-gene profile
 - ii. SKY92, 92-gene profile