

# 5<sup>th</sup> World Hematologists Congress

August 18-19, 2016 London, UK

## Myeloid neoplasms with isochromosome 17q: A distinct entity with unique clinicopathologic and genetic features

Rashmi Kanagal-Shamanna

University of Texas MD Anderson Cancer Center, USA

Isolated isochromosome 17q, i(17q), accounts for less than 1% of myeloid neoplasms that are commonly classified as myelodysplastic/myeloproliferative neoplasms, acute myeloid leukemia (AML), myelodysplastic syndrome (MDS) or myeloproliferative neoplasms (MPN). These cases have distinctive clinicopathologic features with an aggressive clinical course and poor prognosis. However, their molecular mutation profile has not been studied. Here, we present the mutation profile of myeloid neoplasms with isolated i(17q) that included AML, MDS/MPN, MDS and MPN. In addition to the i(17q) as a common finding, these neoplasms had frequent mutations in SRSF2 (55%), SETBP1 (59%), ASXL1 (55%) and NRAS (31%); TET2 and TP53 mutations were rare. Eight of 28 patients (29%) showed concurrent mutations in ASXL1, SRSF2, SETBP1 and RAS. There was a significant association between mutations in SETBP1 and RAS ( $p=0.003$ ). The mutation pattern was independent of the morphologic diagnosis. Sequential mutational analysis of a subset of cases showing evolution from a diploid karyotype to i(17q) showed that SRSF2 and ASXL1 mutations precede the detection of i(17q) whereas SETBP1 mutations are associated with i(17q).

### Biography

Rashmi Kanagal-Shamanna is an Assistant Professor in Hematopathology and Molecular Diagnostics Laboratory at The University of Texas at MD Anderson Cancer Center, USA. She is board certified by the American Board of Pathology in Anatomic and Clinical Pathology, Hematopathology and Molecular Genetic Pathology. She has received her MD from St. John's Medical College, Bangalore, India. She has completed Residency in Anatomic and Clinical Pathology from Henry Ford Hospital, Detroit, Michigan. She has completed her Fellowships in Hematopathology, Advanced Hematopathology as well as Molecular Genetic Pathology at The University of Texas MD Anderson Cancer Center, USA. She is actively involved in bone marrow/lymph node pathology and clinical development and reporting of next generation sequencing-based assays for cancers. Her primary research interest includes application of novel molecular techniques to understand tumor pathogenesis and ultimately guide therapy. She has numerous highly cited peer-reviewed publications in her field and has given talks at many national meetings.

[rkanagal@mdanderson.org](mailto:rkanagal@mdanderson.org)

### Notes: