

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

May 08-09, 2017 Barcelona, Spain

## Mutational analysis in hematological diseases

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**Background & Aim:** Mutation analysis testing and targeting selected regions of multiple genes to facilitate diagnosis, classification and selection of therapy in patients with hematological diseases including acute myeloid leukemia (AML), myeloproliferative neoplasms (MPN), and beta thalassemia was reviewed.

**Method:** Genetic examinations were carried out by polymerase chain reaction using specific primers to amplify regions of interest. The amplified sequences were then determined by direct sequencing (Sanger sequencing) and quantitative real time PCR.

**Results & Discussion:** This study analyzed JAK2 mutations (exons 12–15) in 1811 patients tested between 2010 and 2013. 271 patients (16%) were positive for JAK2 mutations with median patient age of 54 years. In agreement with previous reports, 262 patients (96.7%) were positive for the JAK2 p.V617F mutation. Non-p.V617F JAK2 mutations were detected in the remaining nine (3.3%) patients. 87 patients (84%) were diagnosed with MPN, 40.2% with PV, 25.2% with ET, 12.6% with MF, 19.5% with unclassifiable MPN and 2.2% with CML. A variety of AML-specific mutations were screened in bone marrow samples of 100 adult and pediatric AML patients diagnosed between 2012–2014 showed 45% positivity for different kind of gene mutations. However, the frequency of these different mutations in AML is lower in our population compared to previously published studies from different international centers. In a total of 123 CML patients analyzed, 25 (20%) were positive for 11 different mutations across the ABL1 kinase domain mutations (11 patients had T315I, three patients with Y253H, two patients with E255K, and two more patients with F317L). 19 different mutations of the HBB gene were identified in all detectable cases (103 patients). Among these mutations c.315+1 G>A, c.118C>T and c.92+5 G>C were detected in majority of cases (66%) with five novel mutations (c.410 G>A, c.-151C>T, c.68\_74delAAGTTGG, c.316-3C>A, and c.-31 C>T) that are first time reported in Saudi population.

**Conclusions:** The results of this study underscore the importance of screening for genetic mutations in the initial evaluation of patients with suspected MPNs and AMLs or during a workup for relapse and TKI therapy resistance.

## Biography

Salem Khalil completed his MBBS at King Saud University Riyadh in 1984; fellowship of the Royal College of Pathologists of Australasia, Australia in 1992 and; fellowship in Molecular Hematopathology at MD Anderson Cancer Center, Huston, Texas, USA.

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