

# Predictive, Preventive and Personalized Medicine & Molecular Diagnostics

September 01-03, 2015 Valencia, Spain

## *CALR* gene mutations in primary myelo-proliferative syndromes for personalized medicine

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A new mutated cancer gene (*CALR*) has been discovered in December 2013 affecting mainly primary myelo-proliferative syndromes (MPS). The patients with mutated *CALR* have a lower risk of thrombosis and longer overall survival than patients with mutated *jak2* gene and the clinicians continue to face challenges during diagnosis of un-mutated MPS. *CALR* mutations are present in 70 to 80% of un-mutated *Jak2* MPS. The mutations are located on exon 9 of the gene with a loss of KDEL sequence (signal sequence for endoplasmic reticulum) and loss of calcium binding sites with a new basic (instead of acidic) C terminal region. We will present our data: although the first papers were based on new generation sequencing, we wanted to set up a prospective screening based on High Resolution Melting (HRM) followed by Sanger sequencing using the same pair of primers to speed up the process. We tested 180 samples including for suspicion of Essential Thrombocytemia (ET) and primary myelo-fibrosis. We found 21 *CALR* mutations including one deletion which has not been reported so far. Until 12 years ago the diagnosis of MPS patients was only on elimination; *Jak2* and *MPL* mutations, we can today, with *CALR* mutations in addition, find one mutated gene for 90% of the MPS patients which improves the medical care of such patients. The personalized therapeutic approach is under process.

### Biography

Jean Gabert is a Biochemist and Molecular Biologist, who has spent 10 years in the Cancer Centre in Marseilles (France) as Assistant Professor in Haematology after getting his PhD in Immunology. From 1999 to till date, he has been working as a Professor of Biochemistry and Molecular Biology, Head of the department at the University Hospital in Marseilles. His work has always been in the transfer research and availability for patients of new biological tests allowing improving health care. He has been the coordinator of a very successful European network under the Europe against Cancer program (SANCO Commission). He is currently the Head of the molecular platform for cancer in the PACA West region. He has 5 patents and greater than 75 peer review international publications in reputed journals. He recently obtained honour of Executive Master in Health from Science Po Paris (2011).

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