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2010 : March 2010 - Fast Moving Fronts : Ayalew Tefferi Discusses His Work Regarding TET2 Mutations

FAST MOVING FRONTS - 2010

March 2010



Ayalew Tefferi talks with *ScienceWatch.com* and answers a few questions about this month's Fast Moving Fronts paper in the field of Molecular Biology & Genetics.



Article: TET2 mutations and their clinical correlates in polycythemia vera, essential thrombocythemia and myelofibrosis
Authors: **Tefferi, A;**Pardanani, A;Lim, KH;Abdel-Wahab, O;Lasho, TL;Patel, J;Gangat, N;Finke, CM;Schwager, S;Mullally, A;Li, CY;Hanson, CA;Mesa, R;Bernard, O;Delhommeau, F;Vainchenker, W;Gilliland, DG;Levine, RL

Journal: LEUKEMIA, 23 (5): 905-911, MAY 2009

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(addresses have been truncated.)

SW: Why do you think your paper is highly cited?

TET2 mutations are previously undescribed novel mutations whose clinical and pathogenetic relevance is actively being investigated. As such, many investigators are naturally interested in the subject matter and are working on it as well.

SW: Does it describe a new discovery, methodology, or synthesis of knowledge?

TET2 mutations were first described by a group in France led by Olivier Bernard of the Hôpital Necker-Enfants Malades, Laboratoire d'Hématologie in Paris and William Vainchenker of the Institut Gustave Roussy in Villejuif. Their group is quite well known in mutation discoveries and deciphering the biology of myeloid neoplasms.

What our group did in this article was to describe the clinical phenotype of the mutation in a very large group of patients and define its clinical relevance.

"Our group is heavily involved in the science and clinical practice of myeloproliferative neoplasms and myelodysplastic syndromes."

SW: Would you summarize the significance of your paper in layman's terms?

We provided an estimate on the prevalence of TET2 mutations in a spectrum of myeloid malignancies

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and especially in myeloproliferative neoplasms. We then asked the question as to what it meant to the patient who carries such mutations and found that it was not necessarily detrimental to their survival or other disease complications. Others have found that TET2 mutations were good to have in myelodysplastic syndromes.

SW: How did you become involved in this research and were any particular problems encountered along the way?

Our group is heavily involved in the science and clinical practice of myeloproliferative neoplasms and myelodysplastic syndromes. My other paper in the same journal (Tefferi A, Vardiman JW, "Classification and diagnosis of myeloproliferative neoplasms: the 2008 World Health Organization criteria and point-of-care diagnostic algorithms," *Leukemia* 22: 14-22, 2008) has already amassed over 120 citations in less than two years.

SW: Where do you see your research leading in the future?

Our findings need to be validated by others before making any definite conclusions and more laboratory studies are needed to understand the precise pathogenetic contribution of TET2 mutations.

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KEYWORDS: JAK2; MPL; MYELOPROLIFERATIVE; POLYCYTHEMIA; THROMBOCYTHEMIA; MYELOFIBROSIS; TYROSINE KINASE MUTATION; JAK2 EXON-12 MUTATIONS; ACUTE MYELOID-LEUKEMIA; MYELOPROLIFERATIVE-DISORDERS; V617F MUTATION; ACTIVATING MUTATION; ALLELE BURDEN; KIT MUTATION; JAK2V617F; METAPLASIA.

Additional information:

Video: Dr. Ayalew Tefferi, Professor of Hematology and Medicine at Mayo Clinic in Rochester, MN, discusses the findings of his retrospective study (N=176) of survival in patients with primary myelofibrosis younger than 60 years...[→](#)

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