San Ming Wang, Transcriptomics, 2:2
http://dx.doi.org/10.4172/2329-8936.S1.002

OMICS International
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International Conference on
Transcriptomics

July 27-29, 2015 Orlando, USA

Genome-scale mapping of promoter variation through exome sequencing

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Promoter plays essential roles in regulating gene expression. Alteration in promoter sequences can change gene expression, leading to profound impact in evolution, physiology, and disease. Comprehensive analysis of promoter sequences is critical to understand gene expression regulation under biological and pathological conditions. Here we report the development of a method named EPA (Exome-based Promoter Analysis) for the purpose. The method takes the advantage of the random fragmentation used in exome sequencing to obtain the promoter-exon containing sequences. A unique feature of the method is that it preserves the original contents in promoters for sequencing, therefore, allows *de novo* detection of the changes in promoters without a prior knowledge. This overcomes the weakness in hybridization-based methods as they rely on the normal promoter sequences to design probes. Our evaluation of the method in an exome data set generated from human CD4+T helper cells shows that the method provides high sensitivity to detect most of the gene promoters, and high specificity to detect the common, rare and novel variants in the detected promoters, TF binding motifs, and TATA boxes. EPA method provides a simple but effective means for genome-scale promoter study.

Biography

San Ming Wang finished his Master of Medicine 1986 from Shandong Medical University, Jinan, China. He pursued Doctor of Medicine at Genetic Unit, Swiss Institute for Experimental Cancer Research (ISREC)/University of Lausanne, Switzerland. He worked as Assistant Professor from 2004-2009 at Northwestern University. Later he got appointed as Director for Center for Functional Genomics, ENH Research Institute (Now named NorthShore University Health System Re¬search Institute). From October, 2010–till date, he is working as an Associate Professor at Department of Genetics, Cell Biology & Anatomy, University of Nebraska Medical Center, Nebraska. .

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