



Identifying differential expression genes and single nucleotide variations using RNA-seq in metastatic melanoma

D. Liu¹, Z.G. Zhao¹, Z.L. Jiao² and H.J. Li¹

¹Department of Dermatology, PLA General Hospital, Beijing, China

²Department of Dermatology, Clinic of Navy Headquarters, Beijing, China

Corresponding author: H.J. Li

E-mail: HengjinLi@hotmail.com

Genet. Mol. Res. 13 (4): 8153-8162 (2014)

Received August 12, 2013

Accepted January 13, 2014

Published October 7, 2014

DOI <http://dx.doi.org/10.4238/2014.October.7.10>

ABSTRACT. Melanoma is a malignant tumor and one of the most frequent metastatic cancers. This study was conducted to identify differential expression genes (DEGs) and single nucleotide variations (SNVs) in metastatic melanoma. We analyzed microarray data of GSE23056 downloaded from the Gene Expression Omnibus, including two normal samples (skinN1 and skinN2) and 2 metastatic melanoma samples (skinT and lymphT). We not only compared DEGs in metastatic melanoma samples with normal samples (lymphT_skinN and skinT_skinN), but also compared DEGs between two metastatic melanoma types (lymphT_skinT). SNVs were identified by using Burrows-Wheeler Aligner and Cufflinks in metastatic melanoma samples using RNA-seq. Sequence Alignment/Map tools and the ANNOVAR software were used to analyze and annotate SNVs. We identified 18 significantly common DEGs in lymphT_skinN and skinT_skinN and one common gene, *YBX1*, in lymphT_skinN, skinT_skinN, and lymphT_skinT. We identified 49,534, 48,118, 63,812, and 33,096 SNVs in skinN1, skinN2,

skinT, and lymphT, respectively. Twenty-nine SNVs were located in exonic regions of two DEGs, *HLA-B* and *TSPAN10*. SNVs that exist only in tumors were located in *MARVELD1*, *SLC16A3*, and *VAV3*. The DEGs screened in our study are potential biomarkers for metastatic melanoma therapy.

Key words: Metastatic melanoma; Differential expression genes; RNA-seq; Single nucleotide variations