AKT2, AKT3), RB pathway (RB1 and CDK2) and cellular differentiation (K8). The expression profiles were investigated by real-time qPCR in formalin-fixed and paraffin-embedded tissues, and correlated to immunohistochemical-based molecular classes, namely luminal A, luminal B, Her2 + and TN. The study was approved by the Ethical Committee of the University of Trieste. Results and discussions In our cohort lymph node involvement resulted to be related to the contribution of several genes at the primary tumour tissue level. Some of those genes resulted to be more expressed in LN negative BC, such as PIK3B, RB1 and AKT3, while some others were more expressed in LN positive BC, such as HER2 and AKT1. Our results show higher expression levels of PIK3B and AKT3 in less aggressive BC and higher expression levels of AKT1 in more aggressive BC highlighting the complex regulation of that pathway in BC. Shorter cancer specific survival was recorded in patients expressing higher levels of AKT1 and AKT2. Furthermore, better cancer specific survival was recorded in luminal A BC patients expressing higher levels of AKT3 (p=0.005 in LN- and p=0.01 in LN+).

Conclusion By comparing gene expression in lymph node negative and lymph node positive breast cancers, we found that AKT3 is an independent favourable prognostic factor for luminal A BC patients. Our results showed that a high expression level of AKT3, but not AKT1 and AKT2 was associated to better outcome and longer cancer specific patients’ survival in those patients who display the luminal A molecular class irrespective of lymph node involvement.

Case-Case GWAS to Identify Germline Metastasis Risk Variants in Sporadic Colorectal Carcinomas

Introduction Colorectal cancer (CRC) is the third most frequently diagnosed cancer and a leading cause of cancer mortality worldwide. Majority of mortality is due to metastasis to distal organs such as liver or lung. Stage IV CRC patients by definition have distant metastasis. Up to 50% of stage III (with node involvement) and 25% of early stage I/II CRC patients succumb to distal metastasis. Stage III as well as high risk stage II CRC patients are offered adjuvant therapy after surgery but not all patients benefit from the chemotherapy. In this study, we wish to look for genes and pathways that contribute to metastasis in CRC to better understand the mechanisms and to search for potential prognostic markers and therapeutic targets.

Material and methods Samples from 2300 sporadic CRC patients age 50 years or more with no family history of CRC and with known cancer staging and metastasis status were subjected to whole genome microarray analysis using Affymetrix SNP6 array. Metastasis-positive status of the patients is confirmed based on distal organ involvement attributable to primary CRC, either from histopathological report or computed tomography/positron emission tomography (CT/PET) scan.

Results and discussions Metastasis-negative status is confirmed with at least 5 years of follow-up with no distal organ involvement. DNA was extracted from fresh frozen mucosa collected at least 5 cm away from the tumours. Samples with genotyping call rate less than 95% were excluded and population stratification was examined based on principal component analysis. At the single nucleotide polymorphism (SNP) level, SNP with call rate <99% and minor allele frequency <0.01 were excluded. Whole genome Correlation/Trend test was performed by comparing patients with stages I/II that did not metastasize to patients from stage IV as well as stages I, II that succumbed to metastasis.

Conclusion We will describe PROMO’s main capabilities and show how it can be used for analysing TCGA/GDC’s Breast Cancer datasets for tumour subtype detection and biomarker identification, as done for Luminal-A subtypes in Netanely et al. Breast Cancer Research 18:74 (2016).

Conclusion PROMO provides researchers with an extensive array of tools for quick analysis of large multi-omic cancer datasets.

PROMO is freely available for download at http://agct.cs.tau.ac.il/promo

Introduction Modern genomic datasets may include thousands of samples, each measured by several high-throughput technologies and described by extensive clinical information. Analysis and visualisation of such large multi-label multi-omic datasets pose significant challenges not easily met by existing bioinformatic tools. PROMO (Profiler of Multi-Omics data) is an interactive tool, designed to meet these challenges.

Material and methods PROMO provides various data exploratory methods, enables applying clustering analysis on both samples and features and utilising various popular useful statistical tests including survival analysis and enrichment analyses of subject clinical parameters. Special multi-omic integrative features include joint multi-omic sample clustering and identification of inter-omic feature correlation.

Results and discussions We will describe PROMO’s main capabilities and show how it can be used for analysing TCGA/GDC’s Breast Cancer datasets for tumour subtype detection and biomarker identification, as done for Luminal-A subtypes in Netanely et al. Breast Cancer Research 18:74 (2016).

Conclusion PROMO provides researchers with an extensive array of tools for quick analysis of large multi-omic cancer datasets.