

*Ministero dell'Università
e della Ricerca Scientifica*

*Università degli Studi
di Palermo*

**PhD Programme in Experimental and Applied Medical Sciences
and Biotechnology:**

**Genomics and Proteomics applied to Oncological and Endocrine-
Metabolic research**

Graduation year 2014

**“DNA MICROARRAY AND BIOINFORMATICS AS TOOLS
TO IDENTIFY A COMMON MOLECULAR SIGNATURE
SHARED BY HUMAN ANEUPLOID CELLS”**

PhD Thesis by:
Sergio Spatafora

Director of PhD Programme:
Prof.ssa Carla Giordano

Supervisor:
Prof. Aldo Di Leonardo

Abstract

Genomic instability is a hallmark of the majority of human tumors explaining the heterogeneity shown by tumor cells. This phenomenon is often associated with chromosomal instability (CIN) and aneuploidy, a condition in which tumor cells lose or gain chromosomes. Previously, we showed that posttranscriptional silencing by RNAi of pRb(1), DNMT1(2) and MAD2(3) is associated with aneuploidy in cultured human cells reinforcing the idea that there are several roads leading to aneuploidy. In the attempt to understand if a common molecular signature exists that underlies aneuploidy and its tolerance in tumor cells, we did post transcriptional silencing of Rb, MAD2 and DNMT1 in human fibroblasts (IMR90) and analyzed their transcriptome by Microarray analysis. Using GeneSpring and the R-software for statistical analysis we identified a number of differentially expressed genes in the three samples analyzed when compared to the gene expression of the control. Some of the identified genes were differentially expressed simultaneously in at least two out of three samples analyzed. These data were analyzed using freeware bioinformatics software (DAVID, GOrilla) that showed the presence of a significant enrichment of genes involved in several biological process like G1/S transition, mitotic cell cycle, DNA Replication and DNA strand elongation.