Título: DEVELOPING ADVANCED COMPUTING TECHNIQUES IN BIOINFORMATICS AND BIOMEDICAL ENGINEERING. APPLICATION IN DIAGNOSIS AND ASSESSMENT OF SKIN CANCER

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Resumen: The sequencing of the Human Genome has opened a new era of opportunities in the field of Bioinformatics. Now more than ever, the biological knowledge about the human being continues to widen thanks to immersion and research dedication in multiple interdisciplinary fields at different scales: Transcriptomics, Genomics, Metabolomics, Proteomics, etc. Both governmental organizations and different international institutions have made strong economic investments in search of providing their research centers and laboratories with the best possible equipment.

The explosion of the number of experiments carried that have been carried out in these last 2 decades on the

different technologies of sequencing at transcriptomical level (mainly microarrays and RNA-seq) has meant the collection of an enormous amount of information that does not stop growing. Over time, many of these isolated experiments have been shared with the scientific community both publicly and under controlled access. In this sense, the potential of the information stored in such repositories is extremely high and the biological knowledge to be derived may still be an unknown to be revealed. This is due in large part to the fact that the experiments carried out usually have a very reduced number of samples, which implies the extraction of specific conclusions dependent on the characteristics of the cohort of samples analyzed. Bringing together all the multilevel biological information on the same disease, one could collect a much broader and more robust set of data from which to extract more significant results at the biological level and widely supported at the statistical level.

In addition to glimpsing general conclusions about the most prominent biomarkers of a disease, there is the possibility of immersing oneself in the search for more specific biomarkers, taking into account clinical data that offer a much closer approach to the patient and to that what is increasingly demanded in healthcare: the pursued dream of personalized medicine. In this sense, advanced strategies for the efficient integration of information and the selection of reliable biomarkers are increasingly valued and necessary in order to advance the understanding, knowledge and treatment of diseases.

Although the methodological approaches proposed in this thesis can be extrapolated and applied to any type of disease for which there is a relevant number of samples, the research carried out has focused on improving the diagnosis of skin cancer. This cancerous disease is biologically very heterogeneous and its incidence is increasing worldwide, so there is great alarm and social concern. Since cancer is essentially considered a disease on genetic level, all efforts have been made to extract knowledge from two main sources of information at the transcriptomic and genomic levels: gene expression levels and copy number variations.

Besides providing some insights into the most informative biomarkers for knowing skin cancer predisposition, this dissertation opens new opportunities to develop innovative methodological approaches that consider highly heterogeneous data, quantified in multiple omic viewpoints and leading to the establishment of greater awareness and knowledge about the analyzed diseases.