

Current Position: Assistant Research Scientist in the Department of Molecular Physiology and Biophysics at HHMI/University of Iowa

Education: Ph.D. in Molecular Biology (2002) from Universidad Autónoma de Madrid in Spain

Non-scientific Interests: Christian gospel, Hispanic social integration, writing, and tennis

My keen interest in the molecular basis of human disease has driven my scientific career—as highlighted by my work to identify mutated genes involved in alkaptonuria, epilepsy of Lafora, Walker-Warburg Syndrome, and epithelial-derived cancer. These scientific discoveries helped to open up new areas of research for these diseases. During my graduate studies in Spain, under Santiago Rodríguez de Córdoba, I investigated the molecular basis of alkaptonuria, the first human condition identified as a Mendelian trait. I identified and studied the protein that is mutated in this condition, the homogentisate dioxygenase. I continued my research in Holland in the laboratory of Han Brunner where I became interested in the molecular basis of the most severe muscular dystrophy pathology, Walker-Warburg Syndrome. I identified five genes that are mutated in Walker-Warburg Syndrome patients: POMT1, POMT2, Fukutin, FKRP, and LARGE. These five genes code for known or putative glycosyltransferases that share the cell membrane receptor dystroglycan as their main functional target.

My research on congenital muscular dystrophy brought me to the Howard Hughes Institute at the University of Iowa to study under Kevin P. Campbell. Here I learned that dystroglycan is involved not only with muscular dystrophy, but with cancer as well, as loss of dystroglycan functional glycosylation correlates with the grade of epithelial derived cancers. I discovered that LARGE, which is also involved in muscular dystrophy, is repressed at the RNA level in cancer derived epithelial cells and that LARGE re-expression in highly metastatic cell lines revert their invasive phenotype. The details of these findings are published in this paper.

Read Dr. Beltran's article entitled: Loss of α -Dystroglycan Laminin Binding in Epithelium-derived Cancers Is Caused by Silencing of LARGE

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