From Phenotypes to Function

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The focus in the laboratory is to identify and characterize genes that cause human genetic disease. The diseases studied in the laboratory include the craniofacial disorder, Treacher Collins syndrome (TCS) and Wolfram Syndrome (WS), a neurodegenerative disorder whose main symptoms include diabetes insipidus, diabetes mellitus, optic atrophy and deafness. Information from the Human Genome Project has facilitated all aspects of our work. Identification of the gene which causes Wolfram Syndrome includes the use of linkage analysis, gene mapping, cloning, bioinformatics and mutation detection. The gene for TCS has been cloned and characterization of the TCS gene is the current focus in the lab. The protein treacle has homology to a family of nucleolar phosphoproteins but the function of the protein and its role in craniofacial development is still being elucidated. Current studies include the use of yeast-two hybrid screens to identify protein partners for treacle, the use of microarray analysis to identify genes downstream of the protein and the construction of a conditional knockout mouse model to study the TCS during development.