

AAO Foundation Award Final Report

Principal Investigator	George J. Cisneros, DMD, MS
Co-Investigator	Joseph A. Stofko, DMD
Secondary Investigators	Matthew L. Warman, MD
Award Type	Biomedical Research
Project Title	Collagen XI sequence variations in nonsyndromic cleft palate, Robin sequence and micrognathia (This is the title of the published paper. The title you have on file is slightly different. I apologize for not having the exact wording, but the original paper has been packed away in boxes for years).
Project Year	1998
Institution	Montefiore Medical Center, Albert Einstein College of Medicine
Summary/Abstract	<p>My research mentor, Dr. Matthew Warman, in collaboration with researchers from Norway, combined our patients with theirs to create a larger patient pool. The joint paper, "Collagen XI sequence variations in nonsyndromic cleft palate, Robin sequence and micrognathia," was published in the European Journal of Human Genetics, March 2003, Volume 11, Number 3, Pages 265-270.</p> <p>The paper shows that at least 5% of patients with Robin sequence or micrognathia, who do not have obvious syndromic causes for their problems, will nonetheless have genetic alterations in cartilage collagen genes. This is important in explaining why Robin sequence or micrognathia recurs in some families. 1 in 20 patients with a genetic cause of a seemingly non-genetic condition is a high rate. It was always theorized that a genetic component existed in the nonsyndromic patients. This study finally provided the genetic evidence.</p>