

Vieira Lab Newsletter



Lab Meeting Schedule

- **When: April 13th at 12:00**

Where 403 Salk

Topic: Real Time S. Mutans Assay

Presenter: Kathleen Deeley

- **When: April 20th at 12:00**

Topic: Thesis Defense Practice

Presenter: Ariadne Letra

Upcoming Events

- **When: April 27, 3:30-5:00**

Where: American Cleft Palate-Craniofacial Association, (ACPA), Broomfield, CO,

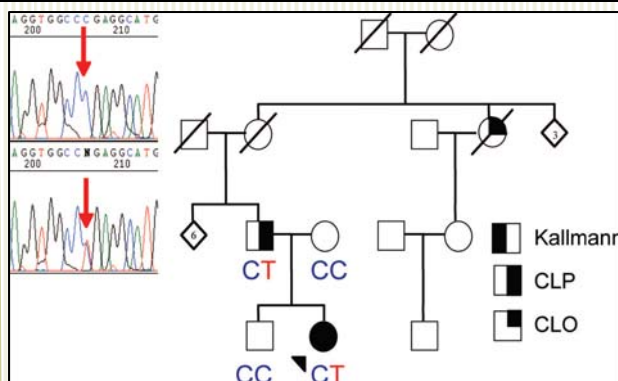
Topic: Concurrent Session XI, Non-Syndromic Orofacial Clefts Panel

Panelists: Alexandre R. Vieira and others



Good Luck Asli!

On behalf of everyone in the Vieira Laboratory, we want to wish Dr. Asli Patir a safe journey back to Turkey. Asli completed two months of research in the Vieira laboratory as a visiting scholar from the University of Istanbul in Turkey. Her research in the Vieira laboratory included the investigation of the genetics of caries. We would like to wish Asli the best of luck in her future endeavors and remind her she always has a home in Pittsburgh.



This pedigree demonstrates mixed phenotypes, with the father presenting as cleft lip and palate only, while his daughter had Kallmann syndrome with cleft lip and palate and tooth agenesis. Both father and daughter were heterozygous for the FGFR1 R609X mutation.

Lab News

This month's newsletter brings a couple of great breakthroughs. Another gene family (the fibroblast growth factor) was shown to contribute to isolated cleft lip and palate in a recent paper published at the Proceedings of the Academy of Sciences USA. Dr. Vieira oversaw the sequencing component of the work. Interestingly, a summer student working with Dr. Vieira in her first week at the lab and doing the first PCR of her life found the mutation that provides the most remarkable finding of the paper (figure above), a nonsense mutation in FGF-receptor1 (FGFR1) in a patient identified as an isolated cleft lip and palate at birth that 25 years later was confirmed as having a syndrome. In another publication, we showed that interferon regulatory factor 6 (IRF6), which we showed associated with isolated cleft lip and palate in 2004, contributes to human tooth agenesis as well. This is a remarkable finding because it suggests that IRF6, that causes Van der Woude syndrome (frequency of 1:100,000 in the population) when mutated, also contributes to isolated cleft lip and palate (frequency of 1:1000) and isolated tooth agenesis (frequency of 1:100). This paper also features FGFR1 and its possible contribution to tooth agenesis.

These papers' references are Riley BM, Mansilla MA, Ma J, Daack-Hirsch S, Maher BS, Raffensperger LM, Russo ET, Vieira AR, Dodé C, Mohammadi M, Marazita ML, Murray JC. *Impaired FGF signaling contributes to cleft lip and palate. PNAS 2007; 104:4512-7* and Vieira AR, Modesto A, Meira R, Barbosa ARS, Lidral AC, Murray JC. *Interferon regulatory factor 6 (IRF6) and fibroblast growth factor 1 (FGFR1) contribute to human tooth agenesis. Am J Med Genet Part A 2007; 143A:538-45.*

Dental Registry and DNA Repository News

As the Dental Registry and DNA Repository (DRDR), has continued to vastly expand its recruitment boundaries within the School of Dental Medicine, it is also quickly approaching the inclusion of its 300th subject, who will donate a saliva sample in the name of facilitating dental research. Exciting things are, indeed, occurring with the DRDR.

On March 5th, the Pittsburgh Tribune Review featured a comprehensive article on the DRDR cleverly entitled, "The Spitting Image of Dental Health." An intriguing video of an enthusiastic subject donating a saliva sample, which greatly complements the article, can be viewed at: http://www.pittsburghlive.com/x/pittsburghtrib/news/s_496142.html Additionally, a student reporter from Penn State University recorded an interview with the study's honest broker, Jill Schaefer, to broadcast over the university's local radio station.

The most significant news of all is the arrival of the first two official requests to use DNA samples and clinical chart information for specific research projects: "Genetic Analysis and Dental Implant Strategies for Dentinogenesis Imperfecta Patients," by Dr. Steven Wendell and Dr. Aysegul Siranli and "PLUNC Proteins as Novel Biomarkers for Inflammatory Dental Diseases," by Dr. Vieira and Dr. Peter Di from the Department of Environmental and Occupational Health. As the second phase of the project, dental research, begins even earlier than anticipated, the DRDR is also very lucky to have 20 eager and talented undergraduate students assisting with the project as well as 4 new dental students. Welcome new students!

To become involved with the DRDR, please contact Jill Schaefer at 412.383.5944 or at jss73@dental.pitt.edu