

A28-1-1339 Experimental Report

Title:

Title: [Genotype-Phenotype Correlation In Human Dental Enamel Affected By Inherited Genetic Disorders](#)

Duration: 08 Nov – 14 Nov 2022

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- The objective of this experiment was to perform a comprehensive texture analysis of dental enamel impacted by amelogenesis imperfecta disease (AI) with the underlying genotype/mutation identified. Investigating how a specific gene mutation disrupts enamel structure, particularly at the nano/cryptographic level, will enhance our comprehension of the protein's role during enamel formation.
- With the assistance of beamline scientists, the subsequent experimental configuration was effectively established to guarantee precise XRD mapping of our dental enamel samples. The detector employed was the Pilatus 1M with dimensions of 981 x 1043 pixels. The wavelength utilized was 0.652548 Å, and the energy was set at 15 keV in multibunch mode, with a beam spot size of 50 µm and a sample-to-detector distance of 250 mm. Counting times ranging between 2 to 5 seconds proved sufficient to attain a robust signal while minimizing noise.
- Number of samples scanned during the beam time: 9 dental samples, comprising 5 teeth affected by AI (KLK4) and 4 corresponding control teeth. The entire crown of each tooth was successfully scanned in specified regions of interest (ROIs), specifically the buccal and lingual halves. This approach maximized beam time utilization and minimized the inclusion of unnecessary dental tissues, focusing solely on enamel. The resolution used in this study was high enough to capture in details texture changes from the enamel surface all the way to the enamel dentine junction.

- The analysis of the enamel Debye rings focused on the 002 reflection, perpendicular to the c-axis of hydroxyapatite crystallites. This allowed us to assess the texture of affected enamel and make comparisons with healthy enamel through azimuthal integration and profile fitting using Matlab software. We deem the experiment successful as we successfully extracted various crystallographic data, including texture magnitude and distribution, crystallite orientation, the presence of multiple crystallite populations, their distribution, and the angular separation between them.
- Our initial data indicates that the KLK4 gene has a greater impact on texture in lingual enamel compared to buccal enamel, and specifically influences the inner enamel more than the outer enamel. This suggests a crucial role for KLK4 during the early maturation stage of enamel formation.
- We observed variations in the wire position in the collected 2D patterns. As a solution, we determined that transposing the data before analysis would rectify the orientation of the 2D patterns.
- We observed a variation in beam intensity that corresponded to the refills, resulting from beam movement or shift. We had experienced the same issue during our previous experiment A28-1-1295. After consulting with the beamline scientists, who thoroughly investigated and devoted considerable time to addressing the issue, we collectively concluded that during the running of experiments whenever low intensity data was collected, we would run an align motor and align slits commands to restore beam position. For data collected already and showing low intensity then normalizing the data to the photodiode could effectively mitigate these fluctuations.

Despite the drift issues, we can conclude that the beam was optimized for our experiment aims; we would like to extend our sincere appreciation to Dr Laurence Bouchenoire and Dr Oier Bikondoa for their help during the beam time.