Abstract

The purpose of my project is to study the evolution and functional impact of genetic variation of the salivary human gene, mucin-7 (MUC7), which has been previously associated with susceptibility to asthma. I present data pertaining to my polymerase chain reaction based DNA amplification experiments and visualization of DNA fragment size using agarose gel electrophoresis. My results reveal the extraordinary size variation of the MUC7 gene within and among human and nonhuman primate species. Our results also revealed a highly divergent haplotype in humans observed only in African samples. Collectively, our results suggest that MUC7 genetic variation has been evolving under strong and differential adaptive forces and may shed light onto susceptibility to asthma in humans.

Introduction

Mucin-7 (MUC7) is a biomedically pertinent and evolutionarily extraordinary human protein. MUC7 is a highly glycosylated protein synthesized in epithelial cells (Kirkbride, Bolscher et al. 2001). MUC7 is an unusually small mucin, sharing no sequence homology with any other mucin protein (Dekkker, Rossen et al. 2002; Kawasake, Suzuki et al. 2004). MUC7 functions in a protective manner by preventing drying-out by providing lubrication and consequently helps chewing, speech and swallowing (Kirkbride, Bolscher et al. 2001). The central domain of this glycoprotein contains repetitive peptide domains, which contain a large number of potential sites for O-linked glycosylation (Kirkbride, Bolscher et al. 2001).

Genomic location, organization and exonic copy number variation of MUC7 gene in human reference genome.

Methods

Polymerase Chain Reaction (PCR): A technique used to amplify segments of DNA

Gel Electrophoresis: A method used to separate macromolecules based on size and charge

Results

Through our analysis, we found that the MUC7 repeats at copy number levels are significantly divergent among mammals as compared to other exonic tandem repeats in the genome (p<0.01).

Conclusion

With the unique copy number variation diversity, we believe that this gene evolved under pressures from frequency dependent balancing or diversifying selection

References

[References list]

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