

HEALTH RESEARCH ABSTRACT SUBMISSIONS

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College *	College of Medicine
Department *	Molecular and Cellular Biology
Title of Research *	Correlations between SNP genotype and FGFR2 expression within intron 2 of FGFR2
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Introduction & Purpose *	Background: FGFR2 has been linked to breast cancer and nonsyndromic cleft lip and palate (NS CL/P). Intron 2 of this gene is of great interest for its high regulatory potential and sequence conservation. Interestingly, 2 single nucleotide polymorphisms (SNPs) are associated with breast cancer and we also confirmed SNPs in intron 2 are associated with NS CL/P. Objective: To identify SNPs within intron 2 where genotype correlates with FGFR2 gene expression. We will use this data to investigate regions of intron 2 to identify functional variation relevant to NS CL/P.
Experimental Design *	Methods: Foreskins were anonymously obtained from healthy, term newborn males. DNA and RNA were extracted from each foreskin. We genotyped 22 SNPs using DNA from 2 sets of foreskin tissues using Taqman assays. FGFR2 expression was measured using qRT-PCR. The differences in gene expression levels among the three genotype groups were evaluated using one-way ANOVA.
Results *	Results: 115 foreskin samples collected in 2008 demonstrated significant SNPs within intron 2 (rs11200014 p=0.02, rs2981578 p=0.04). 156 foreskin samples from 2009 did not replicate the 2008 findings.
Conclusions *	Conclusions: SNPs within intron 2 demonstrated significant differences in FGFR2 expression based on genotype. We are currently pooling expression data from both years to identify overall trends in a larger number of samples. This study helped narrow the region of intron 2 for further analysis including allele dosage and sequencing analysis.
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