HEALTH RESEARCH ABSTRACT SUBMISSIONS #42

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Title of Research *	Linkage Analysis of Preterm Delivery
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Introduction & Purpose *	Preterm delivery (PTD) is a major public health issue accounting for 3 million deaths worldwide each year and is a complex disease with a substantial genetic component. While there are specific causes of PTD, such as twins, the majority are due to spontaneous preterm labor. There are a variety of approaches to identifying genes associated with a complex trait. Linkage analysis using candidate genes takes advantage of some of the known biology associated with labor and can identify loci even when there is substantial allelic heterogeneity.
Experimental Design *	Candidate genes were chosen based on physiologic pathways thought to play a role in PTD and prior association study findings. We studied 95 single nucleotide polymorphisms in 28 genes among 266 extended families, including 504 premature individuals forming 266 affected relative pairs. DNA samples (n=1460) from affected and unaffected relatives were genotyped using ABI TaqMan assays. Premature infants and mothers of premature infants were defined as risk cases in independent nonparametric multipoint linkage analyses.
Results *	Analyses with the premature infant as the case identified two genes with significant evidence of linkage (i.e. NPL p-value <0.01): CRHR1 (p-value=0.0002) and CYP2E1 (p-value=0.002). Analyses with the mother of a premature infant as the case identified four genes with significant evidence of linkage: ENPP1 (p-value=0.003), IGFBP3 (p-value=0.006), DHCR7 (p-value=0.009), and TRAF2 (p-value=0.01).
Conclusions *	These findings suggest the involvement of these six genes in the etiology of PTD. Identification of genes involved in the etiology of PTD will allow a marker set to identify at-risk pregnancies.

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