

## HEALTH RESEARCH ABSTRACT SUBMISSIONS

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<b>Name *</b>	Heather Byers
<b>Email *</b>	<a href="mailto:heather-byers@uiowa.edu">heather-byers@uiowa.edu</a>
<b>Educational Level *</b>	Other
<b>If Selected Other</b>	Medical Student (M3)
<b>College *</b>	College of Medicine
<b>Department *</b>	OB/GYN
<b>Title of Research *</b>	Genetic Causes of Persistent Pulmonary Hypertension of the Newborn
<b>Other Authors *</b>	Jon Klien, MD <sup>a</sup> , John Dagle, MD PhD <sup>a</sup> , Jeff Murray, MD <sup>a</sup> , Kristi Borowski, MD <sup>1</sup> <sup>a</sup> UIHC Department of Pediatrics; <sup>1</sup> UIHC, Department of Pediatrics
<b>Introduction &amp; Purpose *</b>	<p>Background: Persistent Pulmonary Hypertension of the Newborn (PPHN) is a serious, idiopathic, and often rapidly progressive condition. If not treated, it can lead to right ventricle failure, arrhythmias and death. Neonates with PPHN are at high risk for asphyxia and can experience significant morbidity from PPHN and its complications.</p> <p>There is evidence to suggest that genetics could also play a role in persistent pulmonary hypertension of the neonate. Our objective was to investigate potential contributing genes to PPHN.</p>
<b>Experimental Design *</b>	<p>Methods: This prospective study was conducted at UIHC Neonatal Intensive Care Unit from 1993 to 2009. To be included, neonates were diagnosed with pulmonary hypertension, born greater than 35 weeks gestation, and without multiple major congenital anomalies or cyanotic heart disease. 33 SNPs in 9 genes were genotyped using TaqMan chemistry. Biostatistical analysis was performed with Transmission Disequilibrium test (TDT).</p>
<b>Results *</b>	<p>Results: We found significance in the corticotropin releasing hormone receptor gene (CRHR1 rs rs4458044, p=0.00009 and rs173365, p=0.02) and corticotropin receptor hormone binding protein (CRHBP rs10062367 p=0.009). We found a trend towards significance in CRHR1 (rs242944, p=0.08), PDE5A (rs7687843, p=0.09 and rs1480931, p=0.07).</p>
<b>Conclusions *</b>	<p>Discussion: CRHR1 binds to corticotropin releasing hormone (CRH) in the anterior pituitary to stimulate the release of ACTH, which increases cortisol secretion. This pathway plays an important role in the body's response to stress. Identifying a genetic mutation with significant association to pulmonary hypertension could give us a better understanding of the biologic mechanisms of PPHN and open doors for future better identification, treatment and prevention of persistent pulmonary hypertension of the neonate.</p>

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