Diaphragmatic Hernia Research & Exploration, Advancing Molecular Science

Status: RECRUITING

Eligibility Criteria

Sex: ALL
Age:
Healthy Volunteers: This study is also accepting healthy volunteers
Inclusion Criteria:

- All individuals affected with a congenital diaphragmatic hernia (CDH), or with a family history of a CDH

Exclusion Criteria:

- Individuals with no personal history of a CDH or family history of a family member affected with congenital diaphragmatic hernia

Conditions & Interventions

Conditions:
Congenital Diaphragmatic Hernia
Keywords:
Congenital Diaphragmatic Hernia (CDH), Genes, Genetic, Genetic testing, exome sequencing, genome sequencing, RNAseq

More Information

Description:
The goal of this study is to identify genes that convey susceptibility to congenital diaphragmatic hernia in humans. The identification of such genes, and examination of their structure and function, will enable a delineation of molecular pathogenesis and, ultimately, prevention or treatment of congenital diaphragmatic hernia. There are many different possible modes of inheritance for congenital anomalies, including autosomal dominant, autosomal recessive, and multifactorial. Multi-factorial inheritance is responsible for many common medical disorders, including hypertension, myocardial infarction, diabetes and cancer. This type of inheritance pattern appears to involve environmental factors as well as a combination of genetic variations that together can predispose to or produce congenital anomalies, such as congenital diaphragmatic hernia. Our study is designed to establish a small, well-defined genetic resource consisting of 1) Nuclear families suitable for linkage analysis by parametric,non-parametric (e.g. sib pairs, TDT) and association techniques, 2) Individuals with congenital diaphragmatic hernia who can be directly screened for allelic variation in candidate genes, and 3) Individuals who can serve as controls (are unaffected by congenital diaphragmatic hernia). Neonates and their families will be collected from homogenous and heterogeneous populations. By characterizing diverse populations, it should be possible to increase the likelihood of demonstration of genetic variation in selected candidate genes that can then be used in association and linkage studies in individual subjects with congenital diaphragmatic hernia.

Contact(s): Trish Burns - trish.burns@cchmc.org
Principal Investigator:
Principal Investigator ID:
Phase:
IRB Number:
System ID: NCT00950118