

GENETICS OF PAH

Familial pulmonary arterial hypertension:

- can occur at any age
- affects men and women
- however women get disease almost 3 times more often than men
- major risk factor, changes called mutations in the bone morphogenetic protein receptor 2 gene (BMP2)

BMP2:

- belongs to a family of genes that controls cell growth and death
- mutations in this gene make family members susceptible to PAH
- about 80% of families tested have mutations in BMP2
- from 9-40 % of "sporadic" or IPAH patients have mutations
- inheritance is considered dominant with incomplete penetrance meaning that not everyone who carries a genetic change will get disease
- something more is needed to cause development of PAH symptoms

The Something more?:

- another change or mutation in a single gene (unlikely)
- a combination of changes in many genes (probable)
- an environmental factor, for example, female hormone-estrogen
- a combination of environmental and genetic factors

Genetic Counseling and Testing:

- counseling is strongly recommended and often required prior to testing
- testing is available at the following clinical laboratories:
 - Columbia University Molecular Biology Laboratory
 - New York, NY
 - Mahesh M Mansukhani, MD , Director
 - 212-305-2546

[LDS Hospital](#)
[Salt Lake City, Utah](#)
[John Carlquist, PhD, Director](#)
[801-408-1028](#)

Vanderbilt University Molecular Genetics Laboratory
Nashville, TN
Cindy Vnencak-Jones, PhD, Director
615-343-9074

Ambry Genetics Corp
Alisa Viejo, CA
James Thompson, MD, PhD, Director
949-900-5517

- to find a genetic counselor in your area go to www.genetests.org

GINA-Genetic Information Non-Discrimination Act

- signed into law May 21, 2008
- forbids employers and insurance companies to deny employment, promotions, and/or health coverage based on genetic information

-H. R. 493 details can be found at the Library of Congress website www.loc.gov, click on Thomas and enter Bill number