National Institute of Environmental Health Sciences

PEGS Personalized Environment & Genes Study

Powerful science for integrating genomic and environmental data to understand human health

Town Hall Forum
March 28, 2023

National Institute of Environmental Health Sciences

PEGS Personalized Environment & Genes Study

Powerful science for integrating genomic and environmental data to understand human health

A brief history…

The EPR has grown up!

Scientific accomplishments

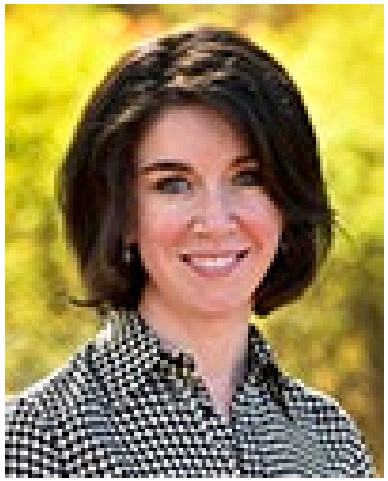
What's on the horizon

Questions

Leadership

Janet Hall, MD –
Co-PI of PEGS
NIEHS Clinical Director, Senior Investigator, Chief of the Clinical Research Branch

Alison Motsinger- Reif, PhD –
Co-PI of PEGS
Associate Investigator, Chief of the Biostatistics and Computational Biology Branch



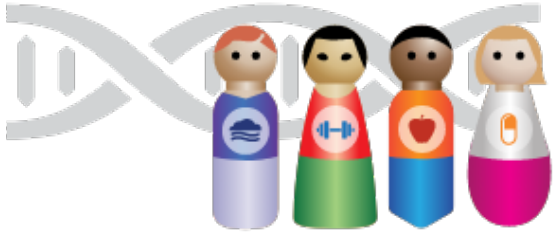
Leadership

David Fargo, PhD –
Director, NIEHS Office of Environmental Science Cyberinfrastructure
PEGS Executive Leadership Committee

Charles Schmitt, PhD *–*
Director, NIEHS Office of Data Science
PEGS Executive Leadership Committee

Internal Advisory Committee
External Advisory Committee

Participant Advisory Committee



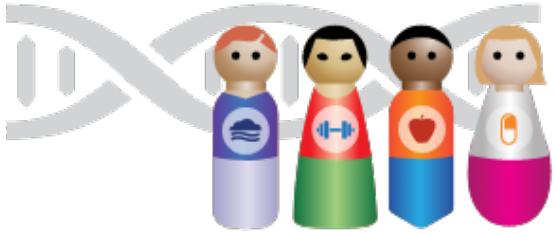
How many total participants have enrolled in PEGS?

A) 5,000 – 10,000

B) 10,000 – 15,000

C) 15,000-20,000

D) 20,000-25,000



How many total participants have enrolled in PEGS?

The answer is: **C) 15,000 - 20,000**

PEGS has enrolled 19,672 participants.

History of PEGS

- Started in 2002, formerly called the Environmental Polymorphisms Registry (EPR)
- Sponsored by the intramural program at National Institute of Environmental Health Sciences (NIEHS) as a long-term project to collect DNA from a diverse group of people in North Carolina
- To date we have enrolled ~ 20,000 participants and are in regular contact with about 50% of these
- Enrollment is ongoing

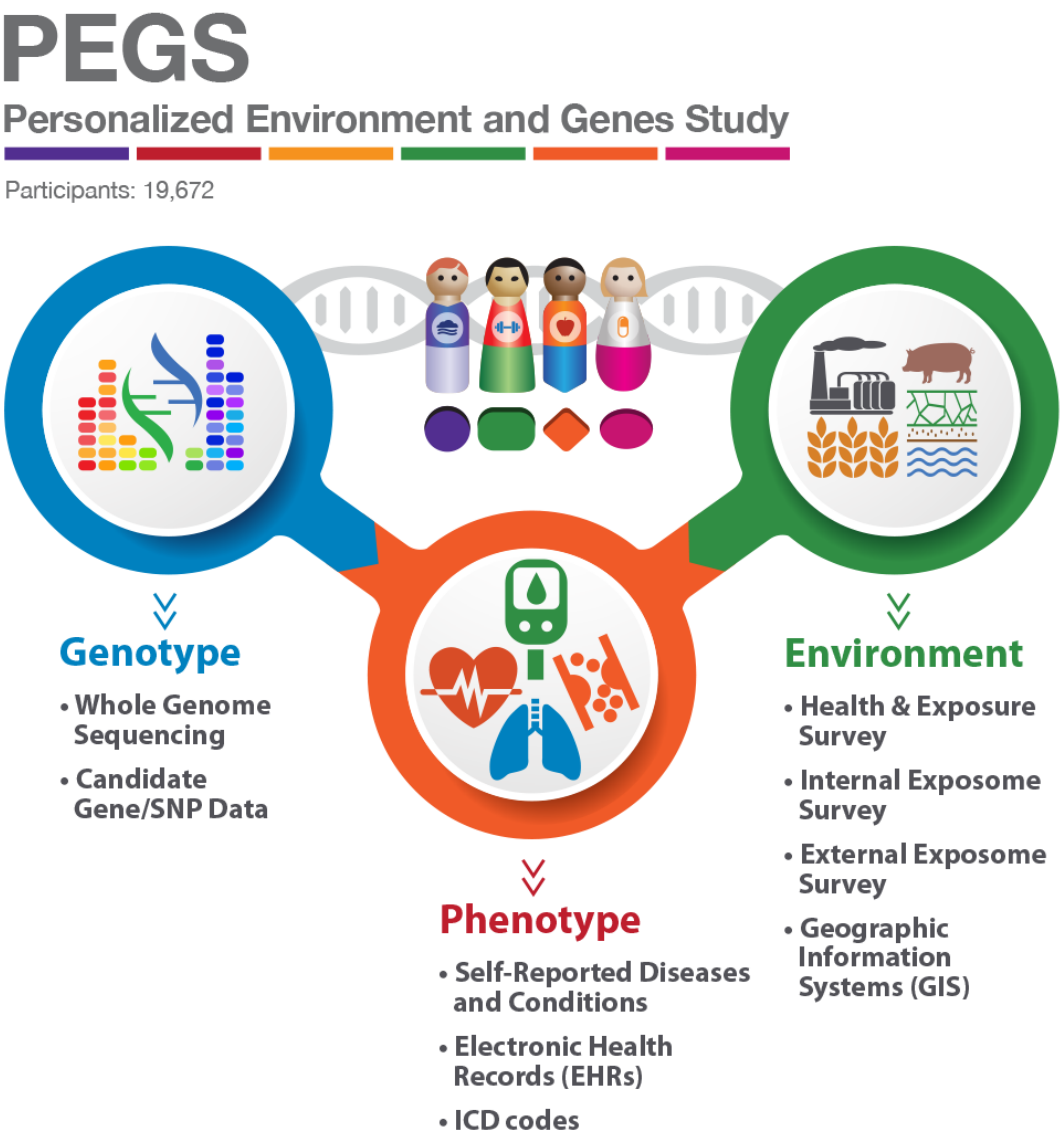
History of PEGS

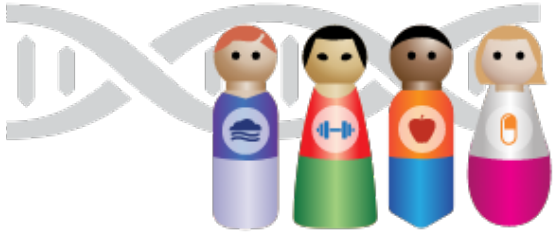
Participants have provided data in various forms

- From 2013-2020, three (3) surveys were used to collect phenotype and exposure data in the cohort
- Information from approximately 9,400 participants was collected
- Types of information collected:
  - General Demographics
  - Family Medical History
  - Lifestyle Factors
  - Occupational Exposure
  - Address histories

- Whole genome sequencing (WGS) for over 4,700 participants

Why the name change?





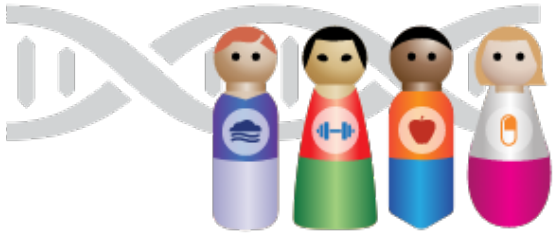
Which is the most common self-reported disease or condition among PEGS participants?

A) Diabetes

B) High Cholesterol

C) Asthma

D) Seasonal Allergies

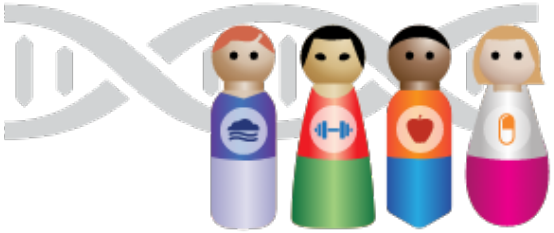


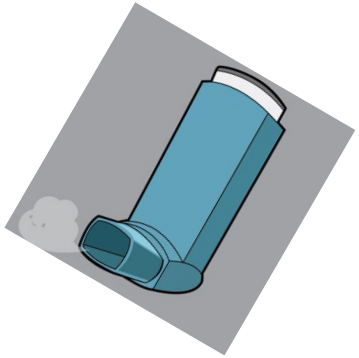
Which is the most common self-reported disease or condition among PEGS participants?

The answer is: **D) Seasonal Allergies**

Seasonal Allergies is the most common self-reported disease or condition among PEGS participants, being reported in 40% of the participants

Scientific Findings -

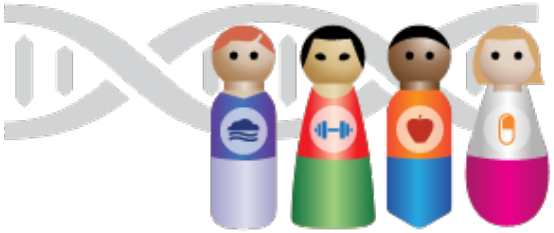




- Distance to roadways increases the risk of asthma and does so to a greater degree in individuals with specific genetic variations

- While there are breaks in our DNA that occur in the skin with normal aging, exposure to UV light dramatically increases this, paving the way for potential malignant changes

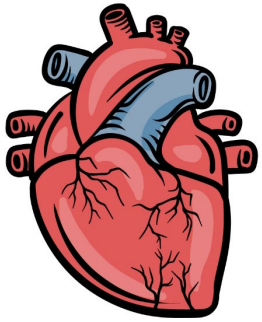
Scientific Findings



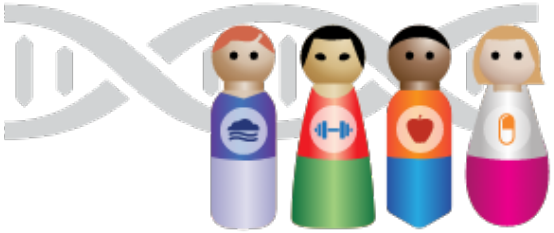
- Recent publications:

- Association of distance to swine concentrated animal feeding operations with immune-mediated diseases: An exploratory gene-environment study (Ayala-Ramirez et al., 2023)

  - Questionnaire-based exposome-wide association studies (ExWAS) reveal expected and novel risk factors associated with cardiovascular outcomes in the Personalized Environment and Genes Study (PEGS) (Lee et al., 2022)



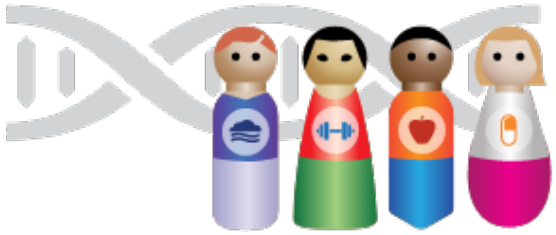
Scientific Findings



- Recent publications:

  - The skin is no barrier to mixtures: Air pollutant mixtures and reported psoriasis or eczema in the Personalized Environment and Genes Study (PEGS) (Lowe et al., 2022)

  - Questionnaire-Based Polyexposure Assessment Outperforms Polygenic Scores for Classification of Type 2 Diabetes in a Multiancestry Cohort (Akhtari et al., 2022)



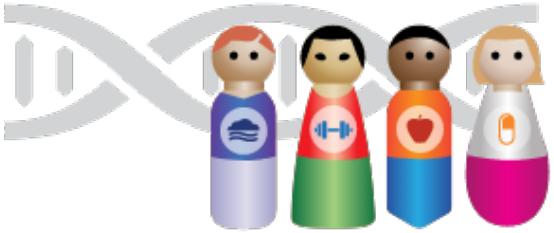
What is the average age for PEGS participants?

A) 18-29

B) 30-44

C) 45-54

D) 55-64



What is the average age for PEGS participants?

The answer is: **C) 45-54**

The average age of participants is 49.7, where the minimum age is 18 and the maximum is 98.

What's on the horizon?

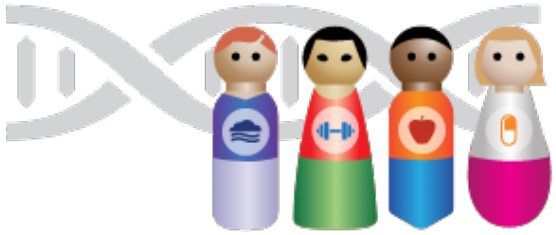
1. Collaborations with UNC and Duke to integrate aspects of **health records** into PEGS
   - Will capture health conditions that may not have been present when you completed the questionnaires
   - Will provide lab tests to confirm the diagnosis

2. **Epigenetics**
   - While our genes are fixed, aspects of our environment can change how our genes function through changes

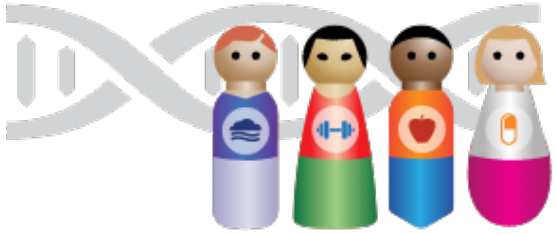
3. **Biospecimens**
   - Collection of additional blood samples, urine, house dust etc to determine the toxins to which we are exposed and how these affect our bodies

Thank you!

By maintaining contact, completing surveys, providing specimens, and updating data PEGS is able to continue to move the science forward and provide you with updates on future events and activities.



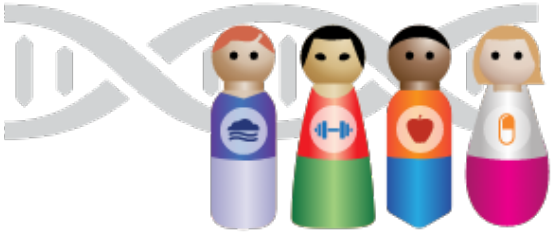
Frequently Asked Questions



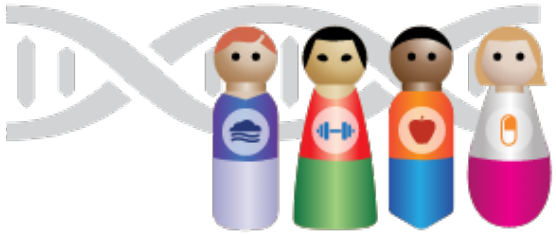
Q: How is my data kept private and/or protected?

- Identifying information (i.e., name, date of birth, etc.) is removed from both data and samples collected.
- Unique codes are assigned to information and a key is created. Keys are kept in a password-protected file on secure servers under layers of security.
  - PEGS PI and Staff are the only personnel with access to the information

Continued…

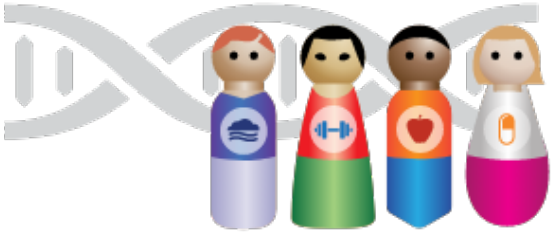


- When granted permission to share your data with other researchers, your data is stored in a controlled access data repository that can only be accessed with approved credentials.
- All who are provided access to the data much sign a confidentiality agreement.
- A Certificate of Confidentially is in place through March 31, 2035.



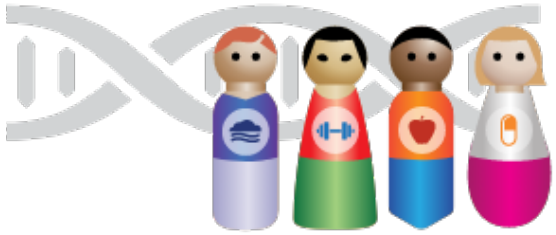
Q: What is being done with my DNA?

- DNA is shared in databases so that approved researchers can “read” your DNA and make determinations on health and disease.
- “Cell Lines” are created to provide an endless source of your cells and DNA for research without the need to collect additional samples.
  - The “cell line” is not used for cloning or to grow artificial organs or organisms and will not be sold or used for any commercial purposes.



Q: What is special about PEGS?

- A diverse cohort – 62% female, 30% Black, range of income and educational levels
- Many different sources of data
  - Environmental cohorts don't have sequencing
  - Genetic cohorts don't have deep environmental sampling
- Participants can be recontacted or called back to the CRU for additional studies



Q: How am I compensated for participating in PEGS?

- PEGS provides compensation based on the sample(s) provided or questionnaires completed.
- The amount compensated varies and is guided by NIH policies and guidelines.

Thank You for Attending!

https://joinastudy.niehs.nih.gov/studies

https://www.niehs.nih.gov/research/clinical/studies/pegs/index.cfm