

# NHGRI Strategic Planning Town Hall

Summary of Prior Meetings & Strategic Planning Events broken into five focus areas and multiple themes within each focus area.



National Human Genome  
Research Institute

The **Forefront**  
of **Genomics**<sup>®</sup>

# Five Focus Areas for NHGRI Strategic Planning

1. Basic Genomics & Genomic Technologies
2. Genomics of Human Health & Disease
3. Genomics in Medicine & Health
4. **Genomics of Data Science**
5. Society, Education, & Engagement



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# Genomics of Data Science: Themes

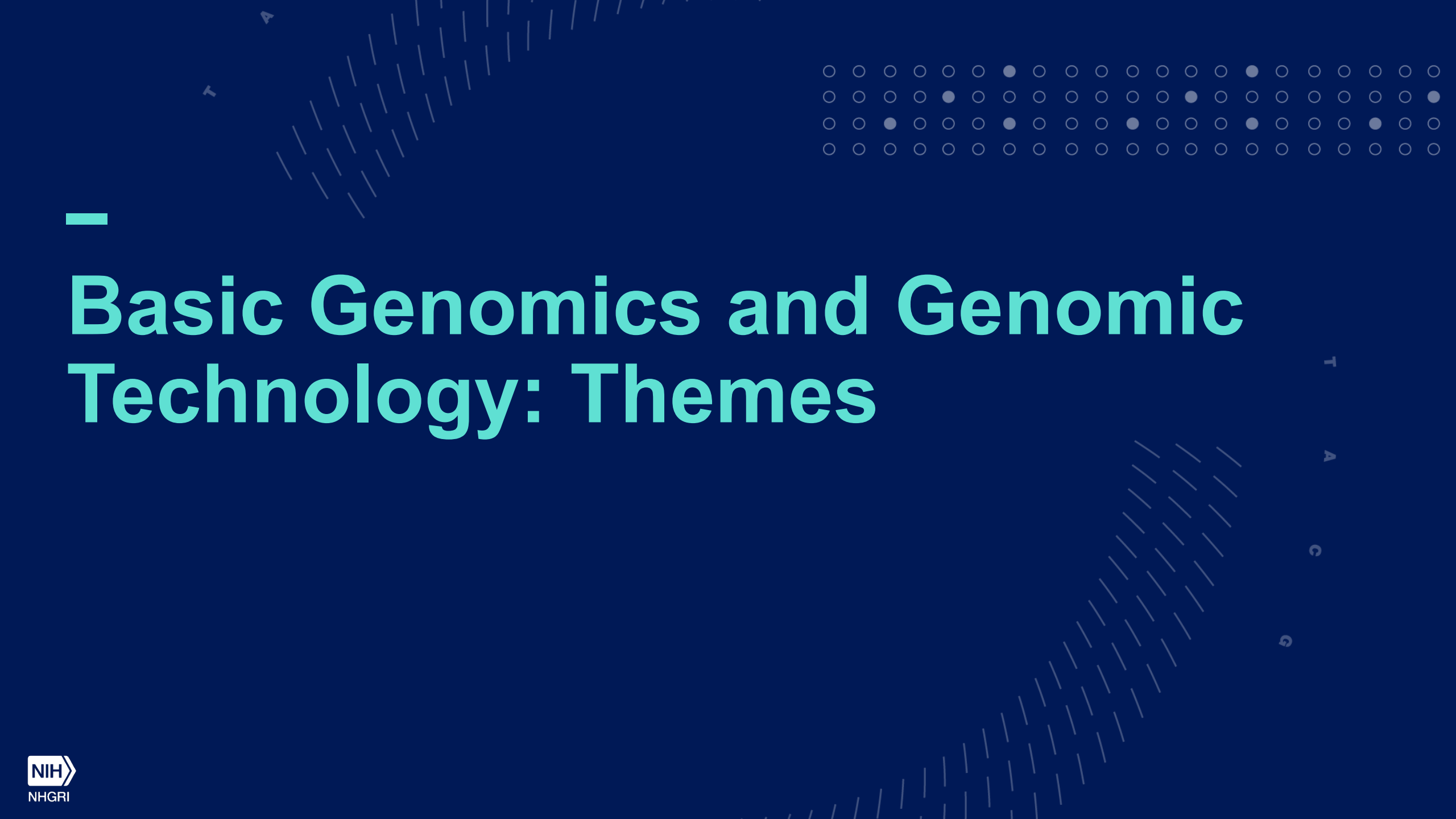
# Focus of Data Science Townhalls

- The next slide summarizes general themes that will be the focus of our townhalls.
- The following slides show a picture of other areas that provide relevant information on the broader goal of NHGRI strategic planning efforts.



# Discussion of Themes

1. Develop computational and statistical methods that facilitate continued advances in genomics
2. Provide leadership for developing and refining genomic data sharing guidelines, policies and best practices
3. Facilitate storing, sharing, and computing on large-scale genomic data
4. Build sustainable genomic information resources
5. Integrate genomic data science into healthcare
6. Ensure that the next generation of genomicists are trained in data science



# Basic Genomics and Genomic Technology: Themes

# Discussion of Themes



- 1. Enable facile, routine generation of whole-genome sequences & transcriptomes and characterization of epigenomes & epitranscriptomes**
- 2. Understand and interpret whole-genome sequences, transcriptomes, epitranscriptomes & epigenomes**
- 3. Establish the role(s) of all genes and regulatory elements in pathways, networks, and phenotypes**
- 4. Use evolutionary and comparative genomic data to markedly advance understanding of genome function**
- 5. Enable facile, routine generation and use of synthetic nucleic acids in genomics research studies**
- 6. Understand and leverage population structure and admixture to facilitate human genetics studies**



**Genomics of Human Health and Disease: Themes**



A C G  
C G T  
A C G

# Discussion of Themes

- 1. Establish the functional consequences of any genomic variant affecting human health and disease**
- 2. Determine the genomic architecture of all human diseases and traits**
- 3. Develop the methods and analyses to support use of non-sequence genomic data for characterizing human health and disease**
- 4. Transform how we assemble sample sets for genomic studies of human disease**
- 5. Commit to systematic inclusion of appropriate ancestral diversity into all large-scale genomic studies and analyses**



# Genomics in Medicine & Health: Themes

# Discussion of Themes

A C G  
C G T

1. Create systems to integrate genomics into everyday clinical and public health practice
2. Improve processes for routine, high value clinical genomic testing
3. Build knowledgebases for predictive genomic medicine
4. Develop and evaluate genomic prevention and therapeutic strategies
5. Ensure that genomics has maximum utility for all members of the public
6. Train healthcare providers to integrate genomics into the clinical workflow



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# Society, Education, & Engagement: Themes

# Discussion of Themes

1. Embrace equity and diversity as core values guiding genomics
2. Meet the expanding educational and workforce needs of educators, health professionals and researchers.
3. Develop and use genomics in accordance with community needs and perspectives
4. Understand whether and how clinical genomics works in specific social and disease contexts
5. Empower well-informed decisions about genomic data use, sharing, and protection, and data stewardship systems that honors those decisions while enabling open science
6. Investigate expanding uses of genomics and their influence on concepts of health, disease, identity, family and community



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