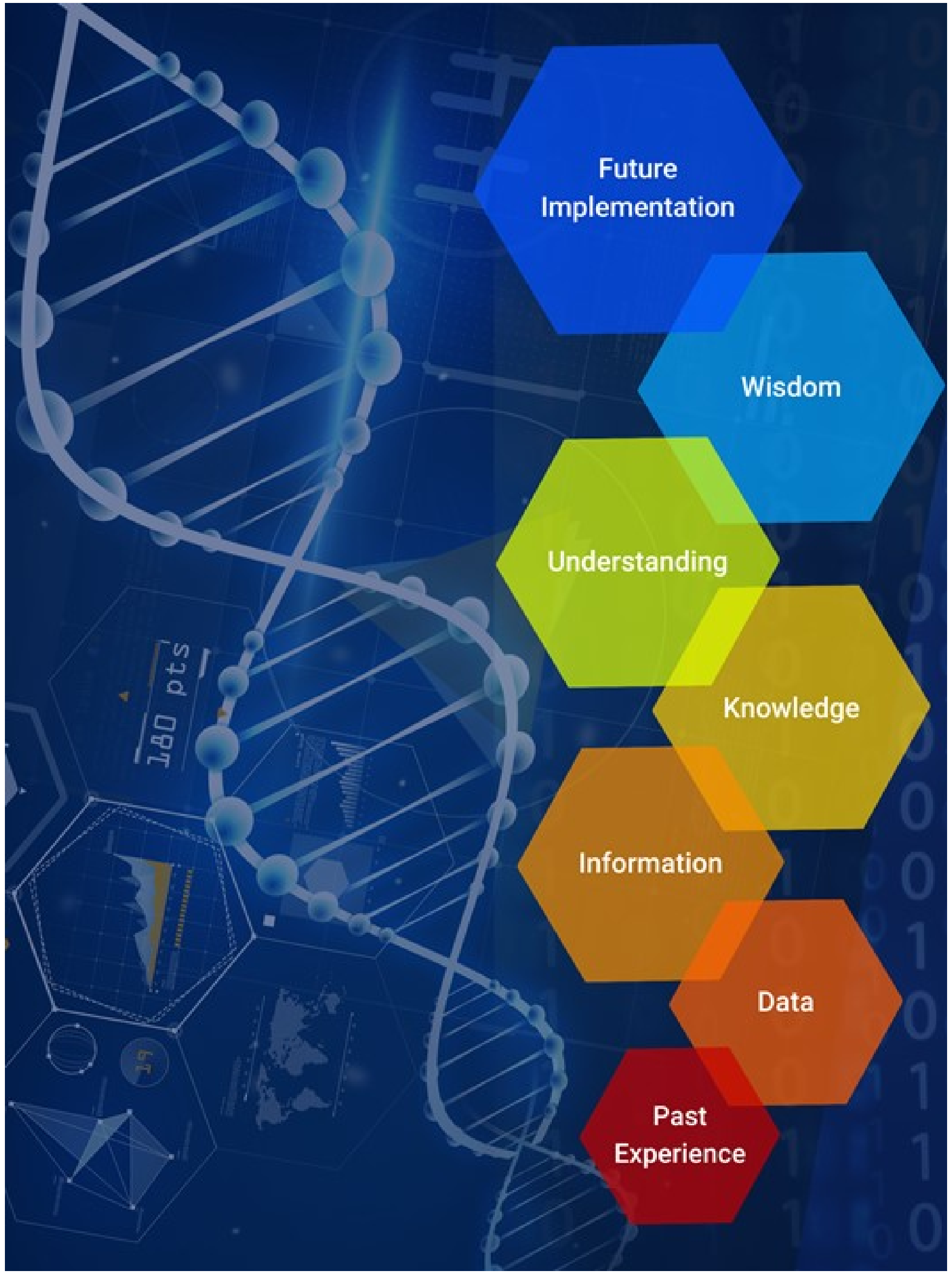
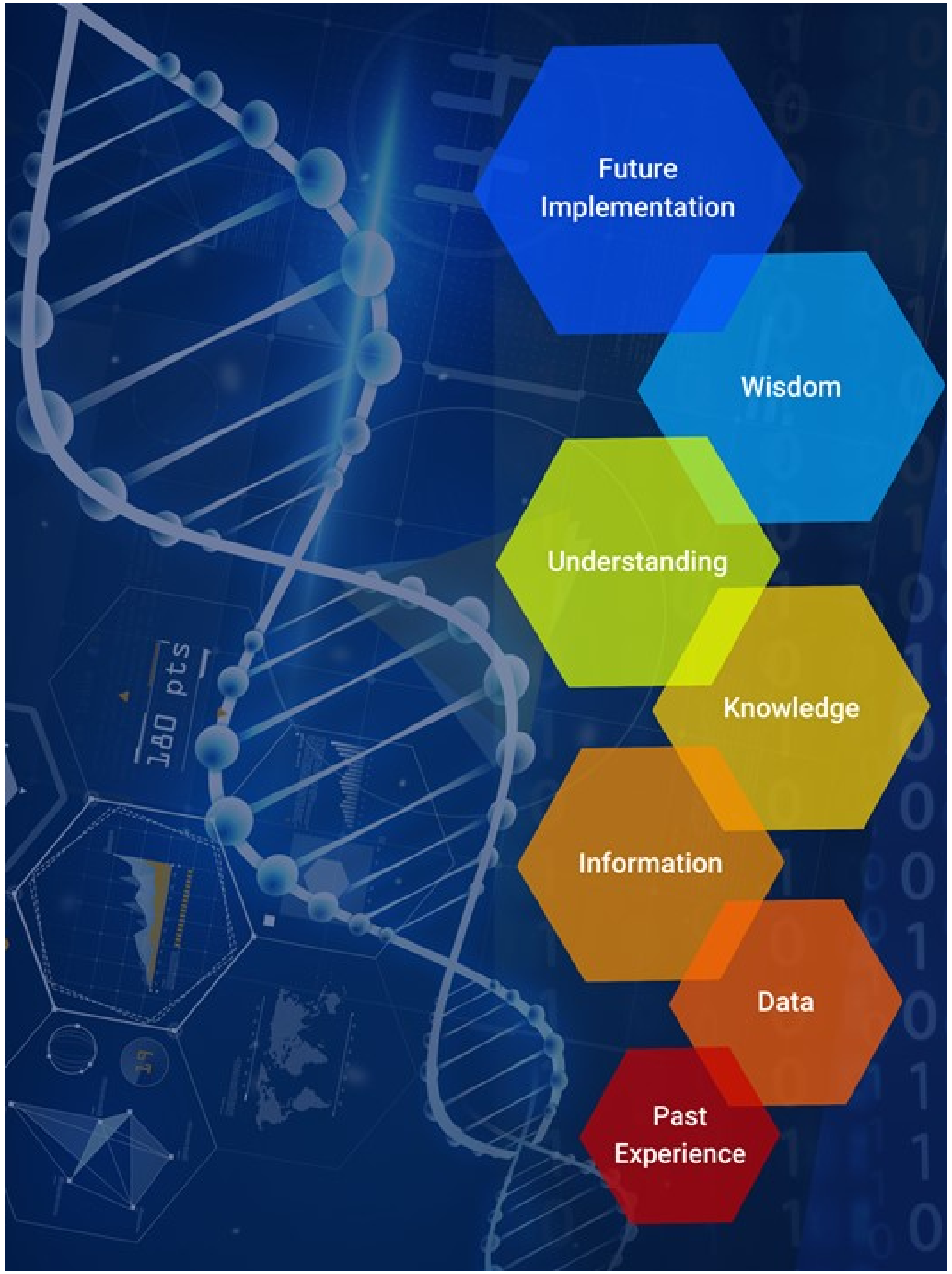
GMXIII Developing a Clinical Genomic Informatics Research Agenda



Goal:

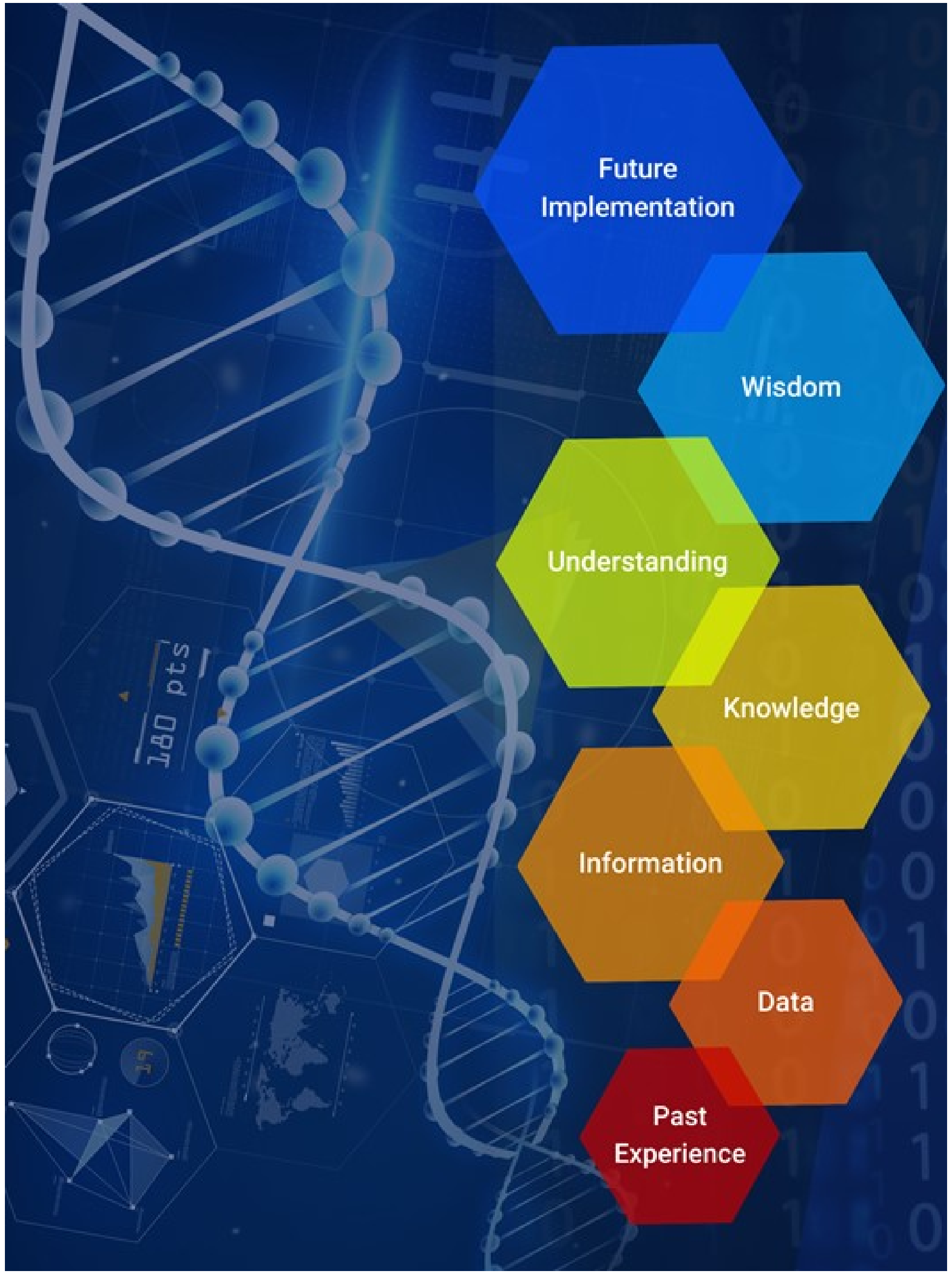
To develop a research strategy on the use of genomic-based clinical informatics resources to improve

the detection, treatment, and reporting of genetic disorders in clinical settings.



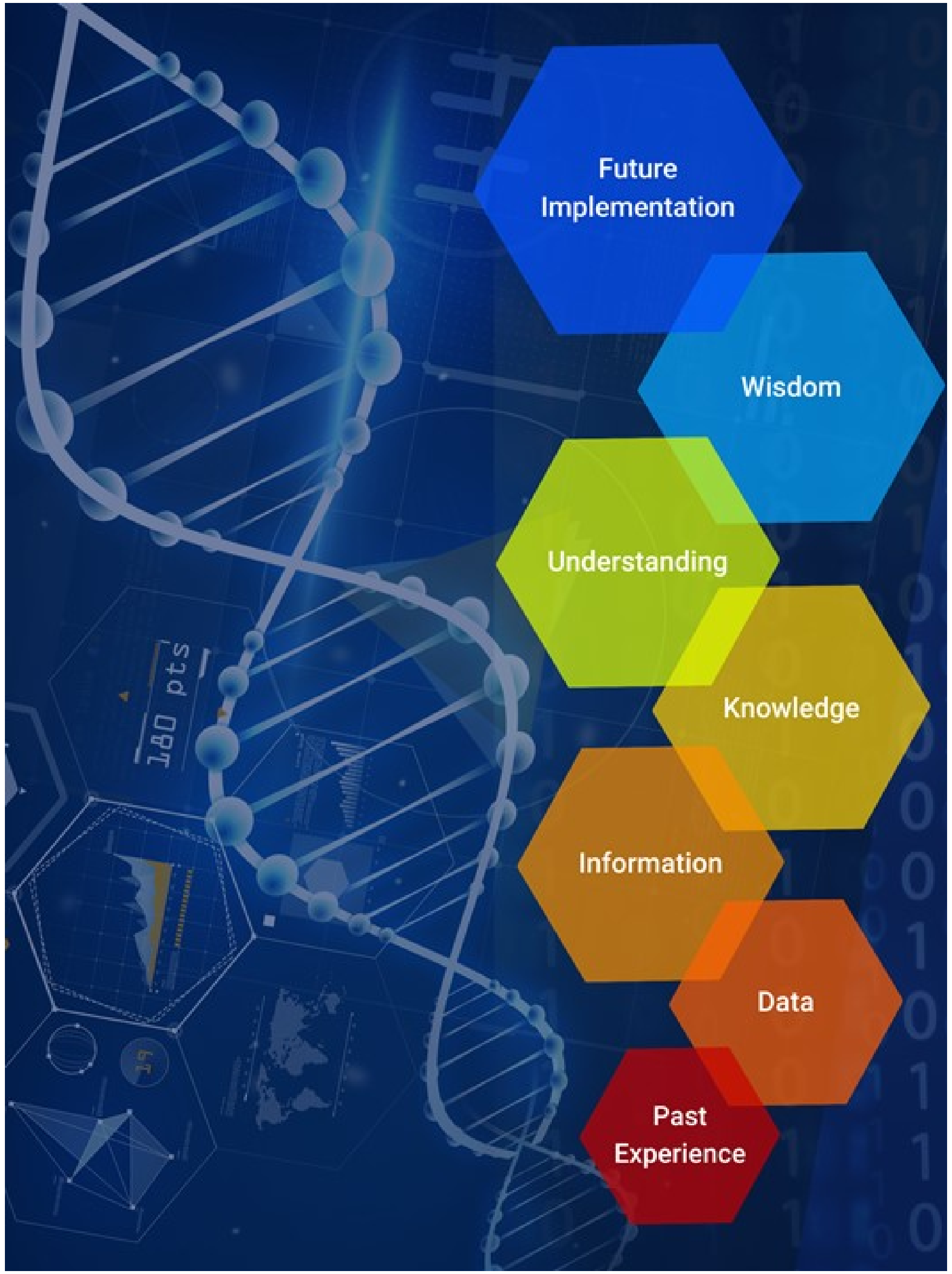
Overarching Themes

- Research should include an implementation science framework
- Value proposition for all stakeholders “patients, research participants, providers, payers, c-suite, a researchers, etc.”
  - support multi stakeholder collaborations
- Need for engagement with patients and partners to develop applications that are under patient control to promote genomic medicine
  - Identify ways to lower barriers related to regulatory processes to promote research in this area
- Recognition of pervasive bias in data, information systems, access, value, and knowledge
- Research needs to account for relevant workflow



Summary--Survey

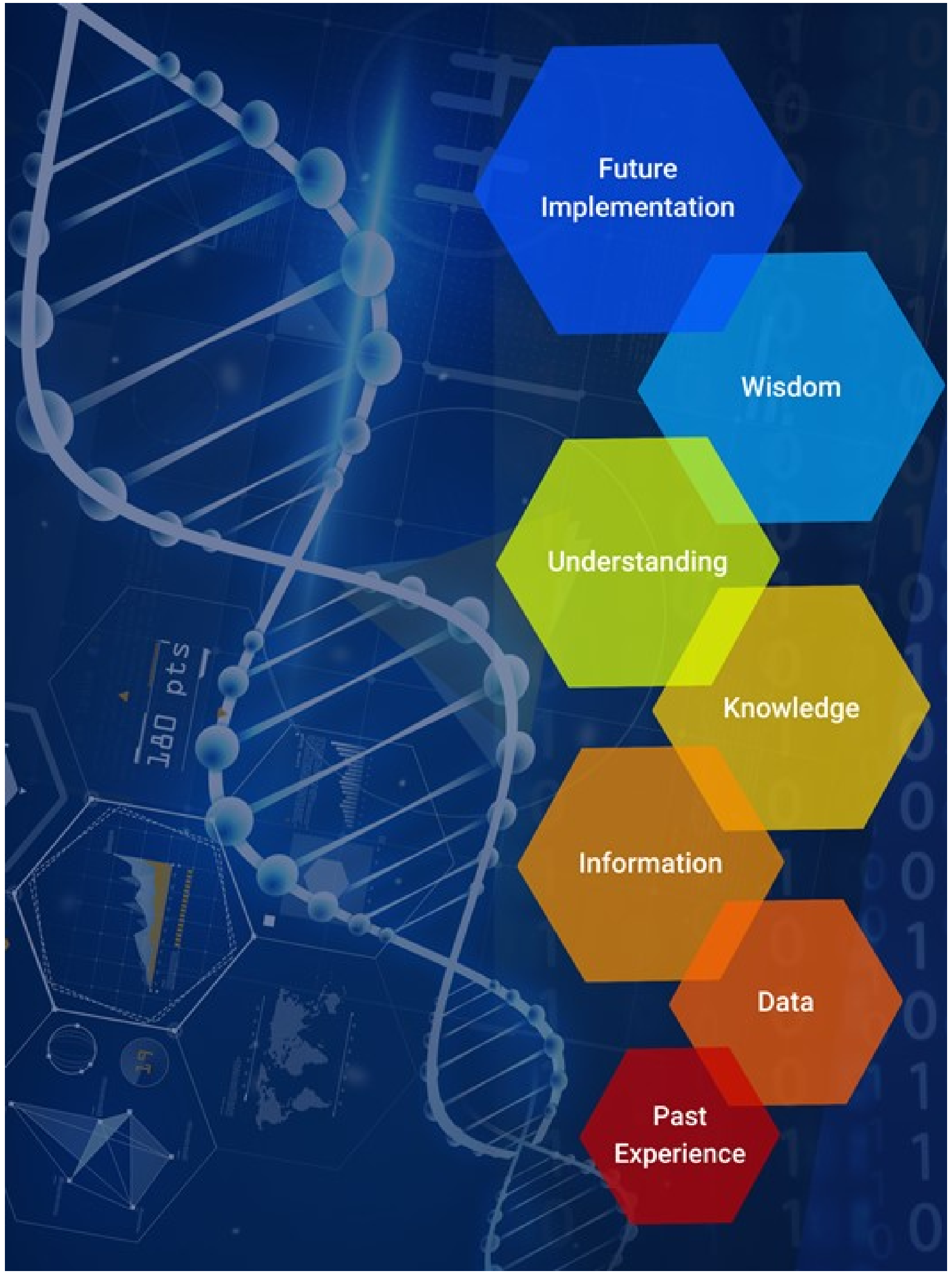
- Key needs from survey
    - Research into methods for integrating analytical interpretations derived by computational models of genomic data into clinical settings are well established (#5)
    - Study ways to ensure that Clinical Decision Support has the ability to incorporate and support multiple genes and clinical info (#8)
- Research is needed that considers preferences in content, preferences, and workflow across a broad range of stakeholders
- Research into the impact of policy and regulation on implementation of Genomic Informatic strategy
- Study of models to support sustainability



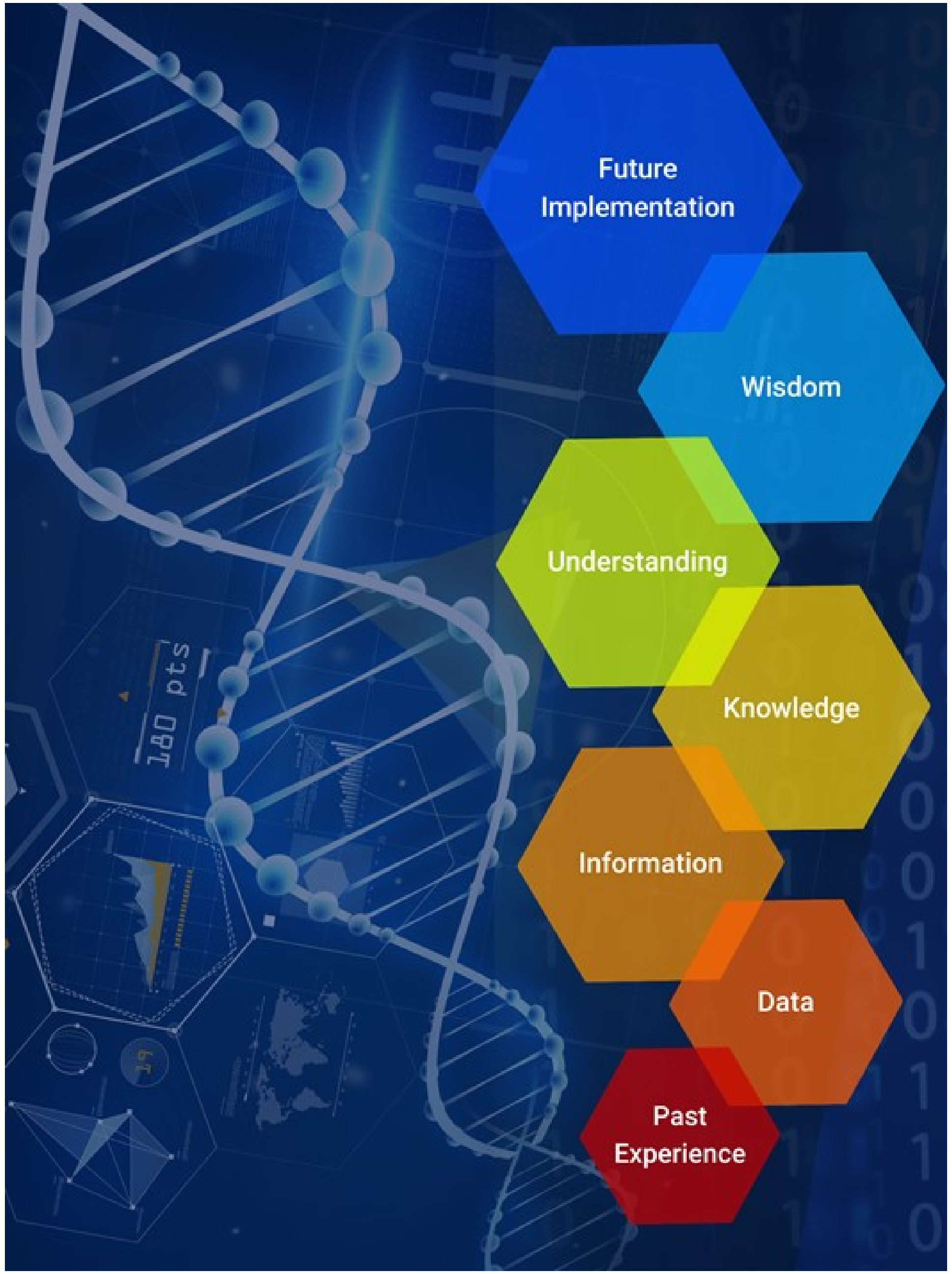
Summary—Session 1

- Must study the inherent biases in data, algorithms, information systems, and implementation. These are multidimensional and include:
  - Race/ethnicity
  - Social determinants of health
  - Urban/rural
  - Academic/non-academic centers
- Need to explore the value proposition which is imbalanced between researchers and participants (more noticeable in underserved populations)

Summary—Session 1 (page 2)



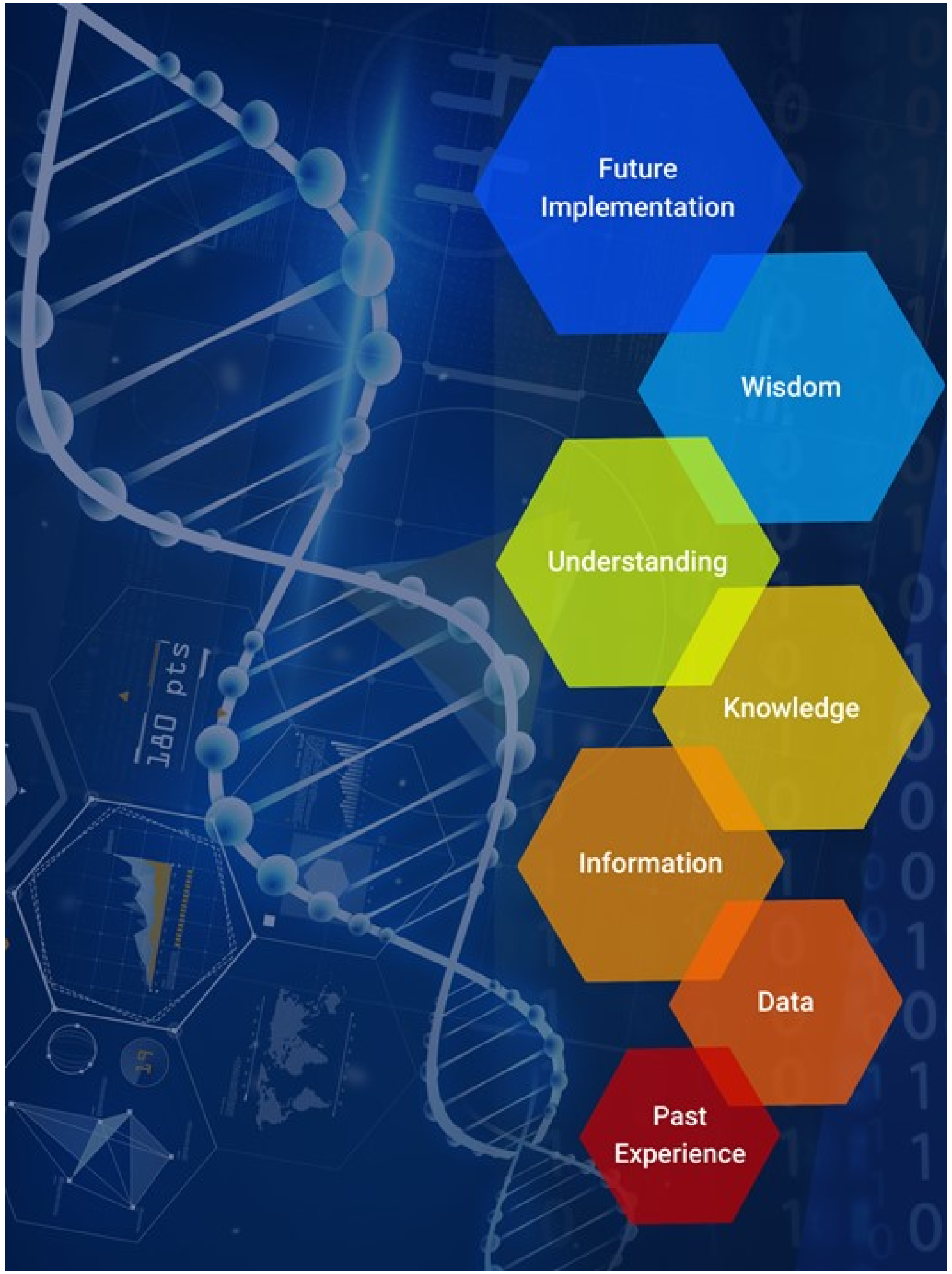
- **Need engagement in research across a broader range of organizations (Casablanca problem)**
    - ○ **Implementation equity**
- **Need innovative research questions and methods to address these inherent biases in a rigorous and systematic way**
- **Outcomes must capture both benefits and harms of gCDS to inform mitigation approaches**



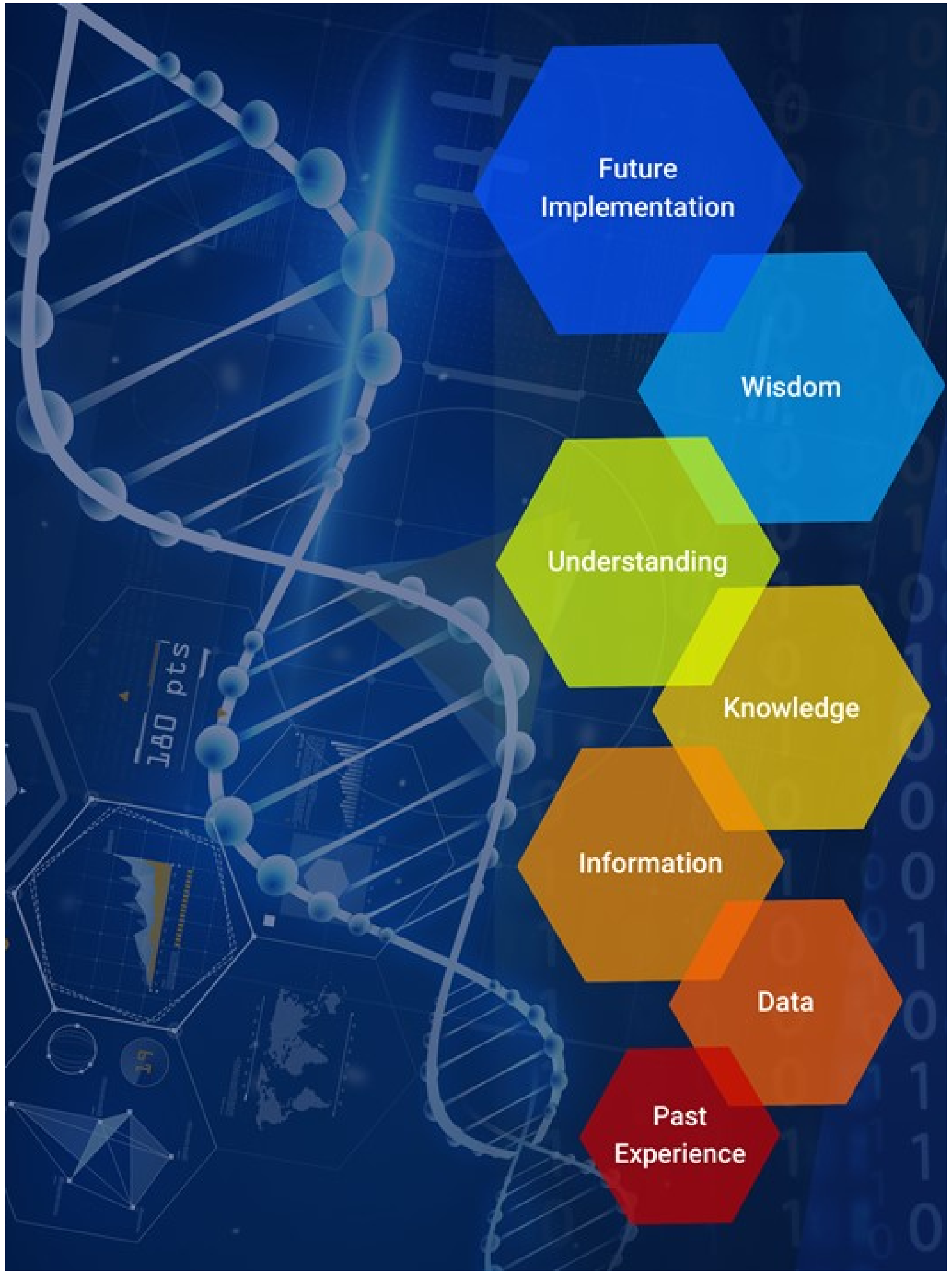
Summary—Session 2

- Use implementation science research methods to improve implementation equity, not just research equity
- Develop a patient-centered research agenda. Example research topics:
    - Authentication, privacy, security of genomic data accessible to patients
    - Recontact of patients absent clinician oversight
    - Knowledge requirements for different stakeholders
    - Innovative enabling platforms for obtaining and returning genetic results
    - Ability of patient-centered focus to reduce bias

Summary—Session 2 (page 2)

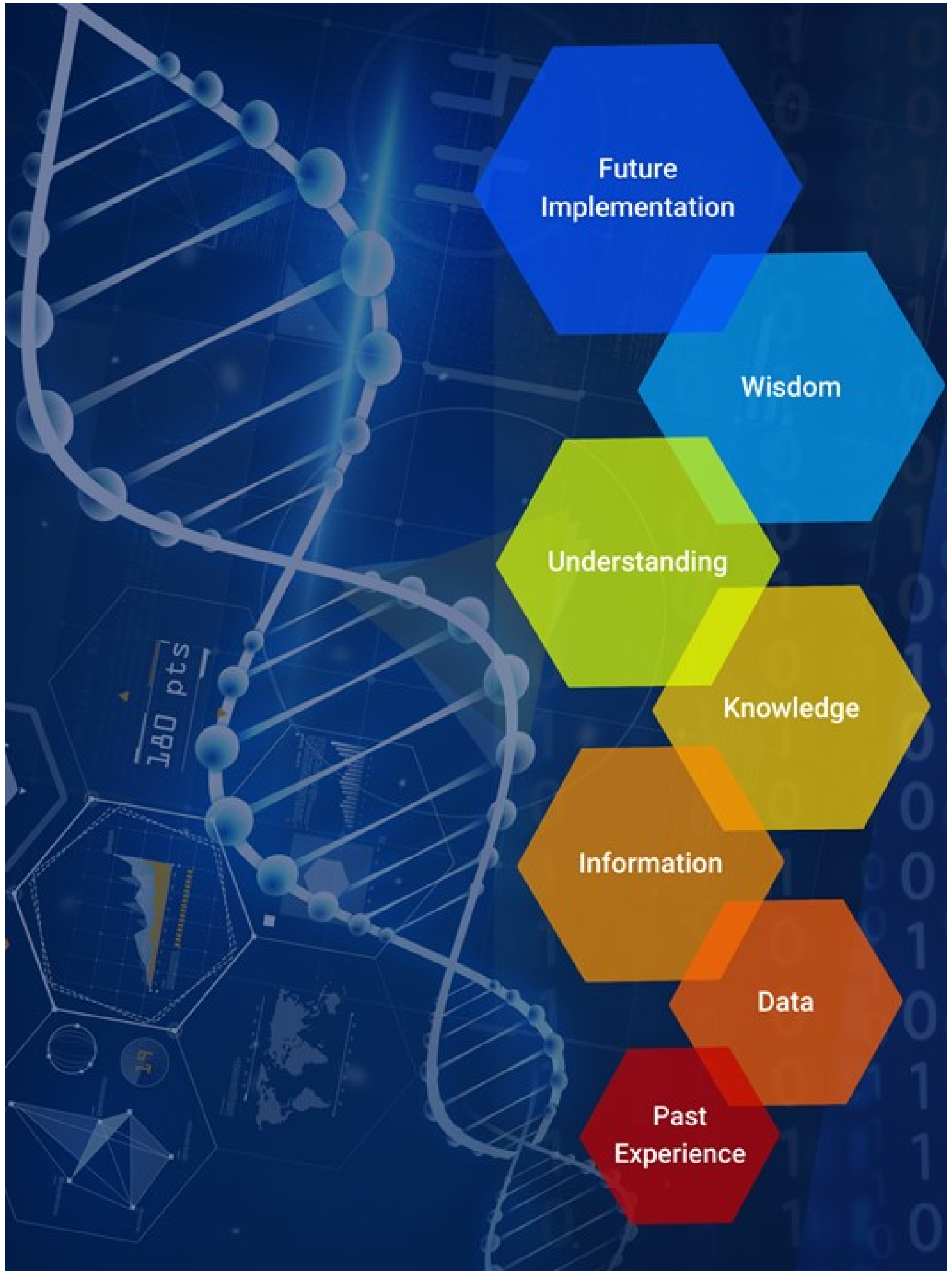


- Need an updated "Business Model" of research attract broad range of stakeholders to participate to understand more about the incentives to implement genomic based clinical informatics resources and tools
- Research into ways to represent genomic information as structured data while minimize manual processes.



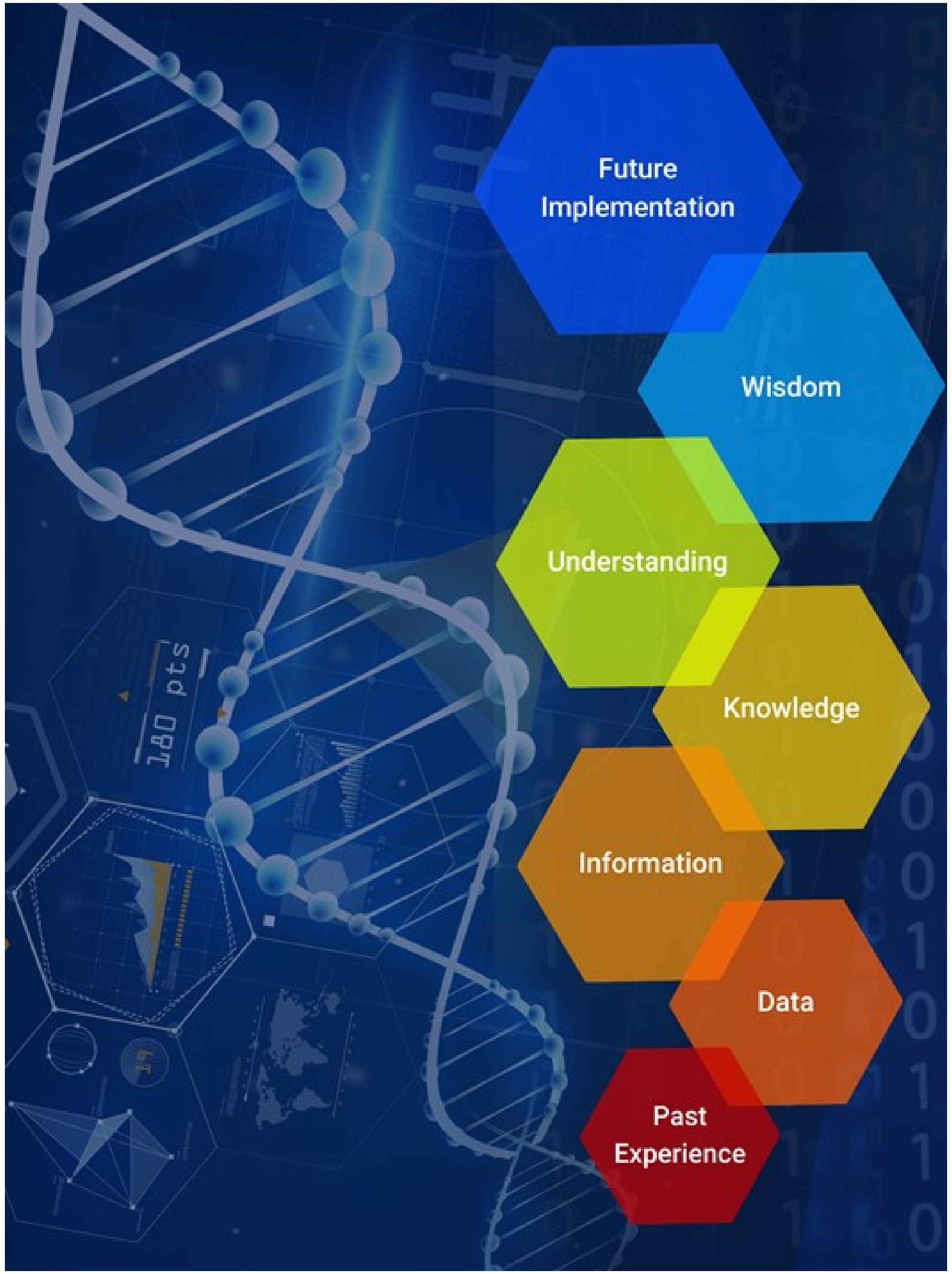
Summary—Session 3

- Informatics research for genomic evidence computing and genomic knowledgebase construction to enable scalable, sharable and computable inferences of genomic knowledge and harmonization of practice guidelines
- Research into novel workflows that diminish burdens for primary care providers, tap into other healthcare workers and engage patients, and don't default to alerts and reminders
- Studies to ensure new technologies don't exacerbate health disparities
- Educational and policy research agenda to reduce barriers and improve knowledge for patients and providers



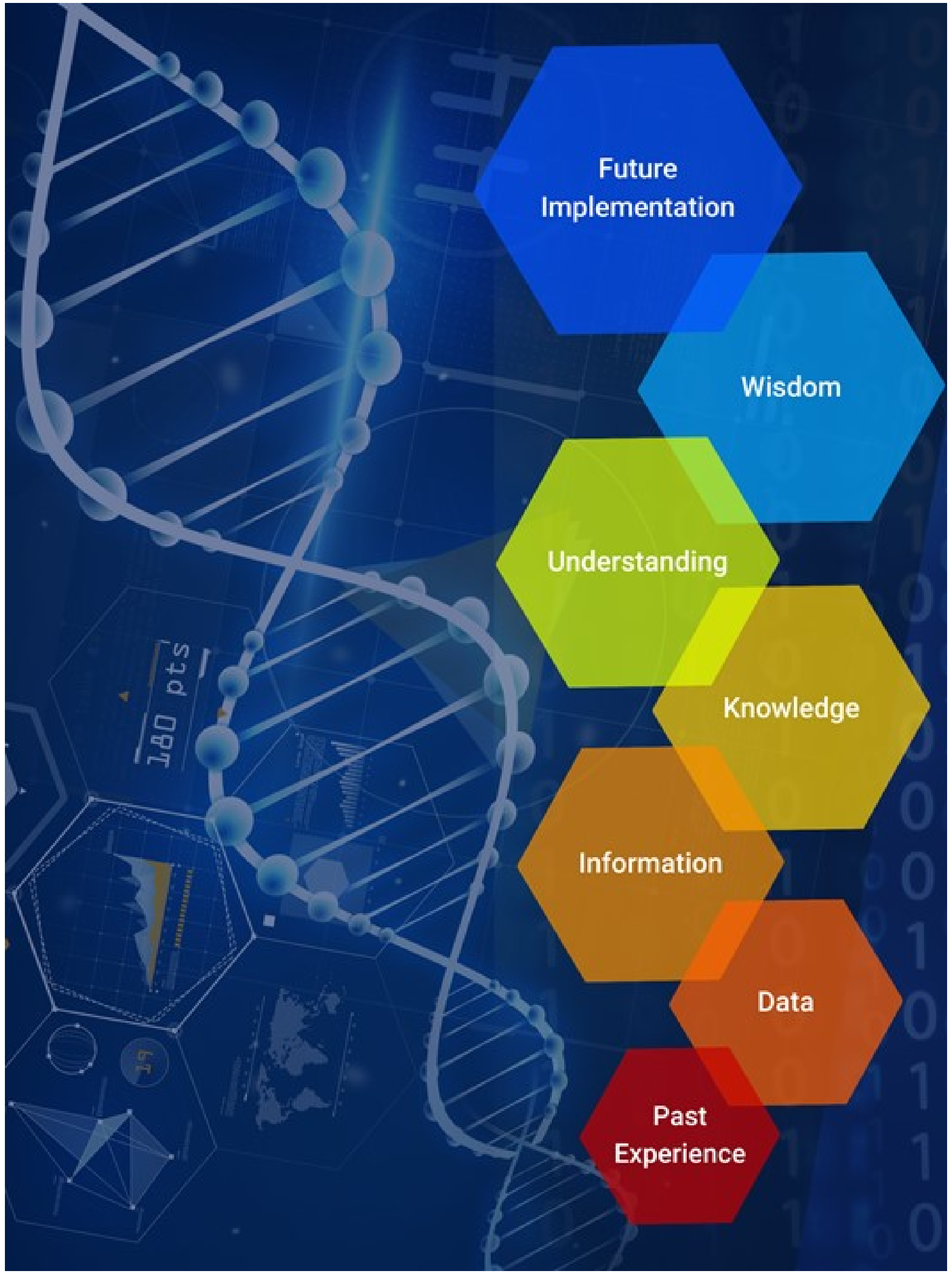
Summary—Session 3 (page 2)

- Sociotechnical Strategies for Success
  - Informatics research for genomic evidence computing and genomic knowledgebase construction to enable scalable, sharable and computable inferences of genomic knowledge and harmonization of practice guidelines
  - Harmonize the interests of multiple stakeholders to facilitate team science and implementation science
  - Incentivize collaborations to foster research on a learning health system for genomics (what is new evidence, who is affected, what needs to be done, etc.)
  - Clinician (and patient)-centered design of workflow support.



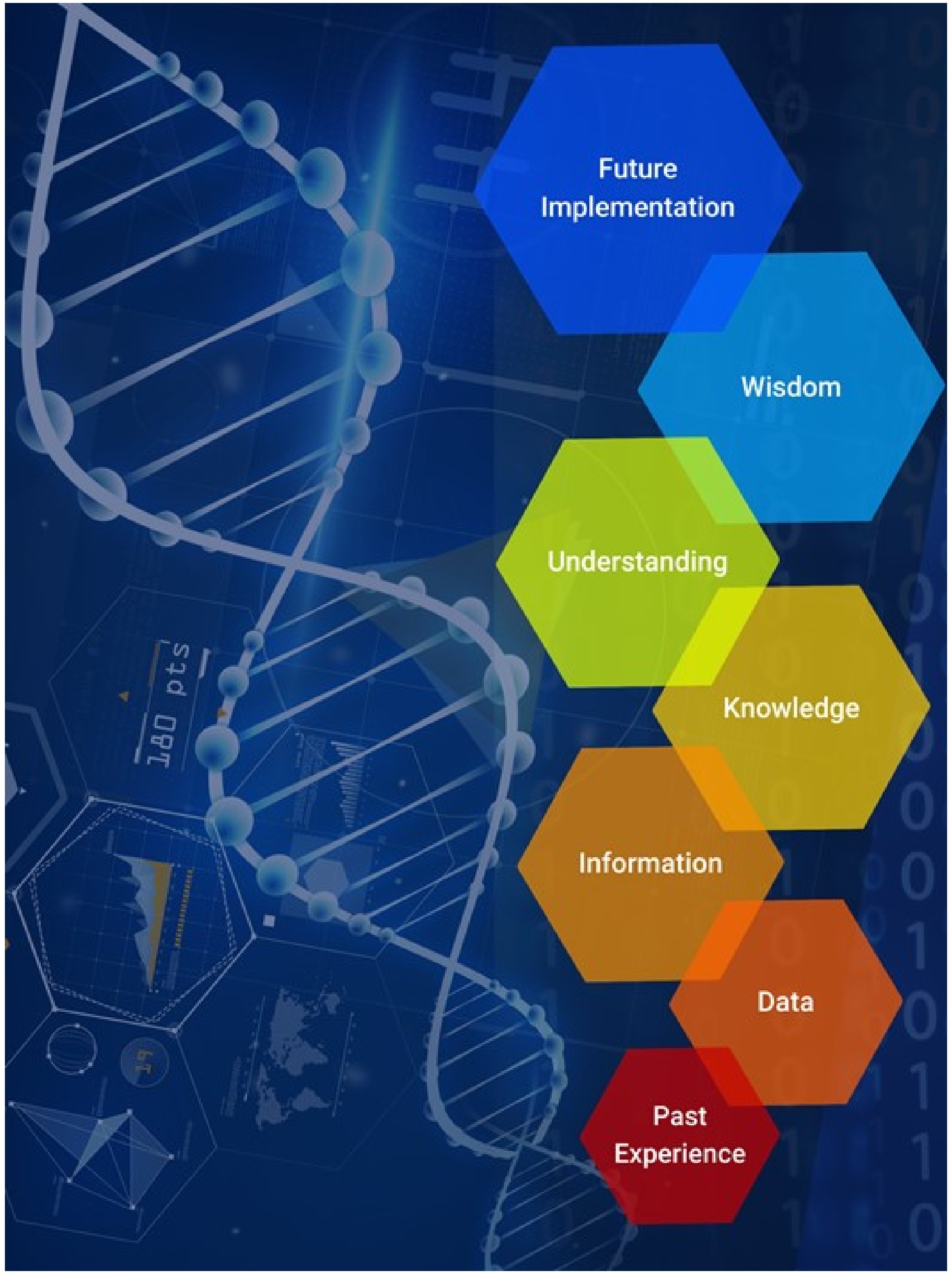
Summary—Session 4

- Research on what constitutes a minimum data for clinical care and research “learn from less data”
- Research at the interface of human cognition and artificial intelligence “how can we take the best of both”
- Include research into explainable AI to promote clinical implementation
- Research into the development and implementation of a common semantic framework to reduce reliance on manual curation



Summary—Session 4

- Research into the development of a responsibility model across EHR for patient access to needed information
- What are 80/20 use cases in genomic informatics



Summary—Session 5

- Research into data interoperability between clinical systems that is focused on the implementation of genomic medicine
- Develop research on specific use-cases to support genomic medicine implementation through informatics
  - Prioritize based off diverse stake holders
- Coordinate and synergize research findings with the broader health IT community (US Core Data for Interoperability, HL7,)
- Facilitate last mile of clinical implementation–identify what’s ready and support implementation research around it


Future Implementation
Wisdom
Understanding
Knowledge
Information
Data
Past Experience
180 pts
19

Thank You!