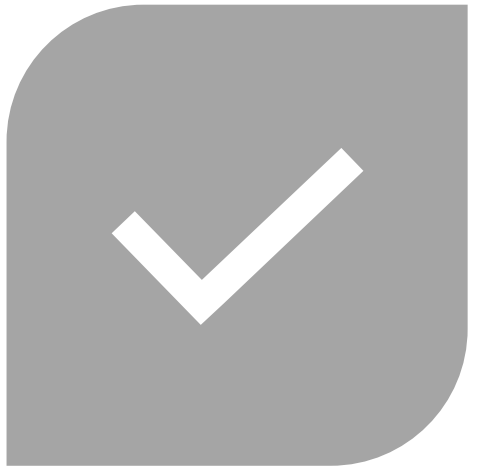
Evaluating the Efficiency, Accuracy, and Impact of Preventive Health Diagnostics through Health Genomics



A STUDY ON THE HEALTH GENOMETER PLAN

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Introduction

- **Overview of Whole Genome Sequencing (WGS)**
  - Emergence in healthcare over the last 20 years
  - High throughput/next generation sequencing methods
  - DNA sequencing and reconstruction

Importance of WGS in Infection Prevention

  - Comparing sequences to assess relatedness
  - Construction of detailed phylogenic trees

- **Significance of WGS**

Understanding Transmission Dynamics

  - Complexity of transmission in healthcare settings
  - Identification of related and unrelated transmission events

Challenges and Philosophical Questions

  - Intensity of infection prevention responses
  - Resource allocation in healthcare-associated vs. non-healthcare-associated transmission

Research Questions

- **RQ1:** How useful is current technology in diagnosing and predicting diseases through health screening?
- **RQ2:** What are the long-term health outcomes for patients who participate in technology-based preventive health check-up programs like the Health Genometer Plan?

Main Objective:

- Evaluate the efficiency, accuracy, and impact of disease detection and prediction technologies.
- Focus on Health Genomics programs.

Purpose:

- Explore benefits, challenges, and ethical considerations.
- Assess patient perceptions, financial impact, and long-term health impact.

Hypothesis

Qualitative Research Design:

- Utilizing secondary data to explore effectiveness, challenges, and implications
- In-depth analysis of existing literature, reports, case studies, and relevant documents

- **Review of Literature**

1. Whole Genome Sequencing in Support of Wellness and Health Maintenance

- Combining clinical and genetic profiles for wellness-focused medical care
- Visualization of risk in subclasses of disease
- Need for training genomic healthcare professionals

2. Disease Diagnosis in Smart Healthcare

- Emerging machine learning algorithms and applications
- Challenges including privacy, pilot studies, and data communication
- Financial viability and environmental sustainability

3. Exploring the Impact and Utility of Genomic Sequencing in CKD

- Better diagnostics and treatments in nephrology
- Integration of genomics into clinical care
- Development of research priorities

Study Design:

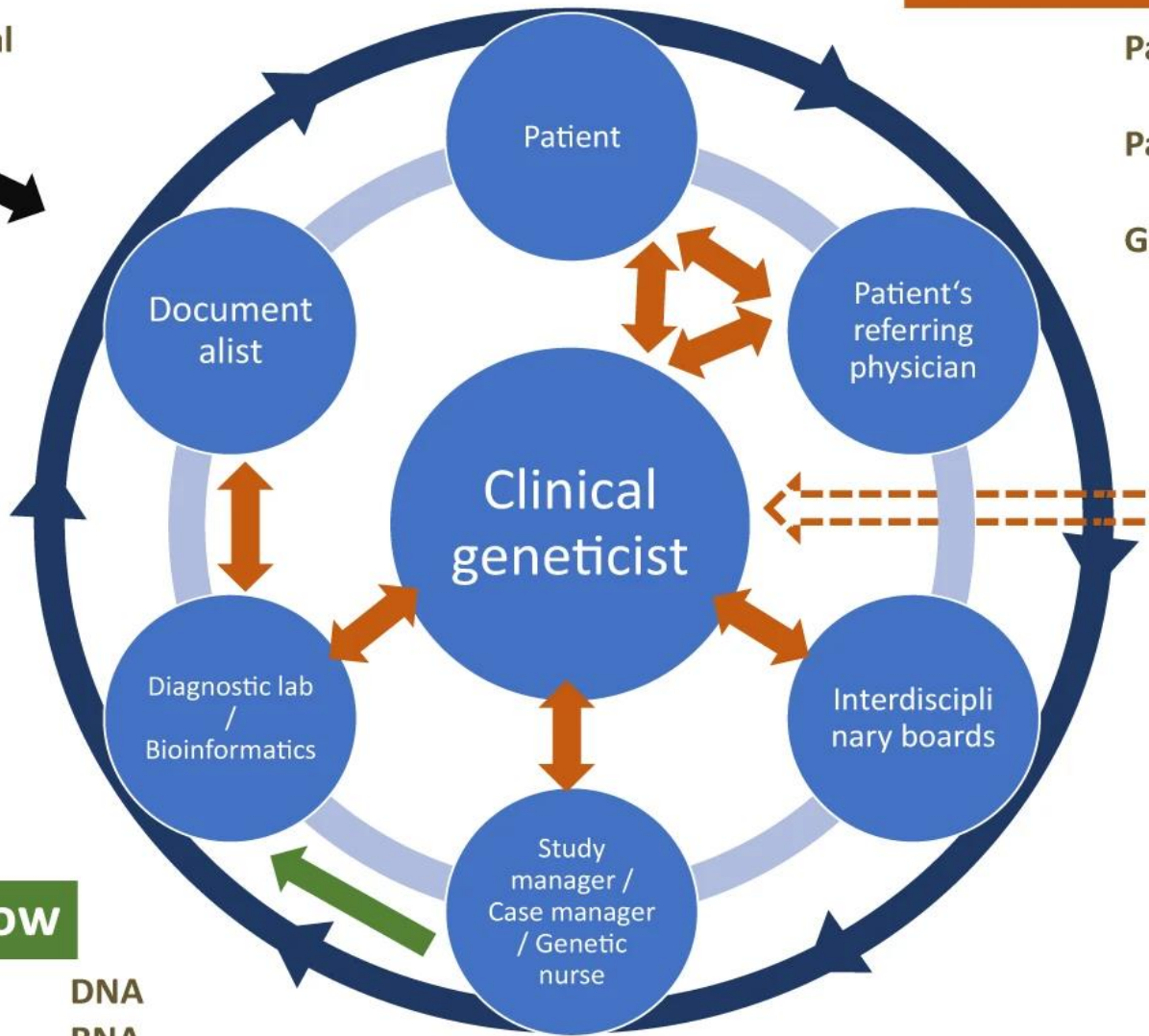
- **Qualitative Study Design**
  - Conducted in a tertiary care hospital in Mumbai
- **Study Population:**
  - Local population of Vikhroli East, Mumbai and nearby areas
- **Duration of Study:** 3 Months
- **Sample Size:** 140

Data flow

Local / National / International
Data access
Data stewardship

Communication flow

Patient's autonomy
(Who will be informed on genomic data?)
Patient's approval
(What data should be reported?)
Global informed consent
(Use of data for research?)



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Clinical decision flow

Reporting
Treatment & Management Decision
Potential research integration
Follow ups

Sample flow

DNA
RNA
cf DNA
Biomarkers

Data Collection and Analysis

- **Data Collection Procedure:**
  - Secondary data collected from the Genometer Plan in the hospital
- **Data Analysis Plan:**
  - Analysis using Excel to determine outcomes of genome sequencing through health check-up
- **Ethical Considerations:**

Adherence to Ethical Principles:

- Informed consent
- Confidentiality
- Anonymity

- **Implications of Research**

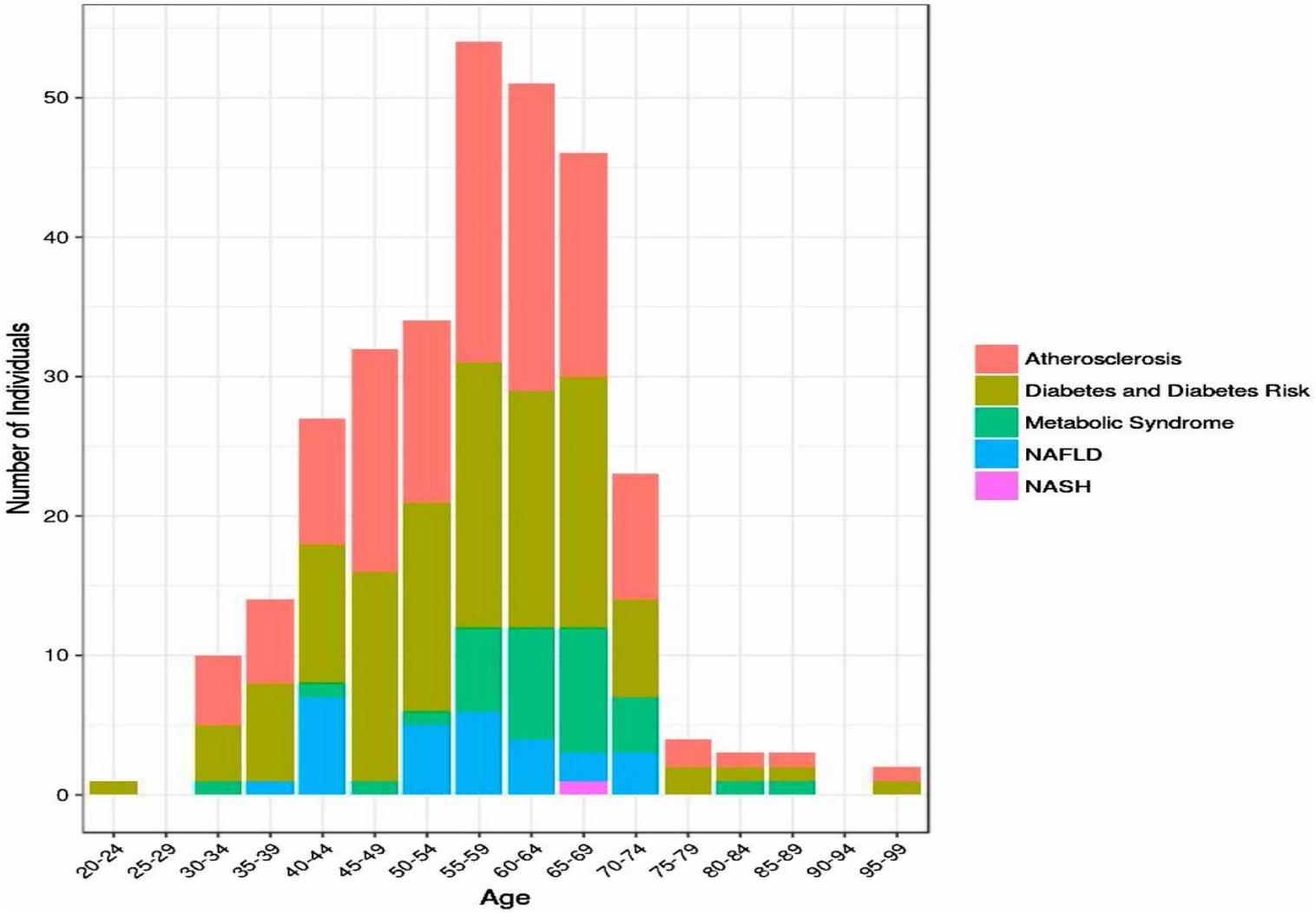
Proactive Health Management:

- Lifelong genetic risks and the opportunity for proactive measures

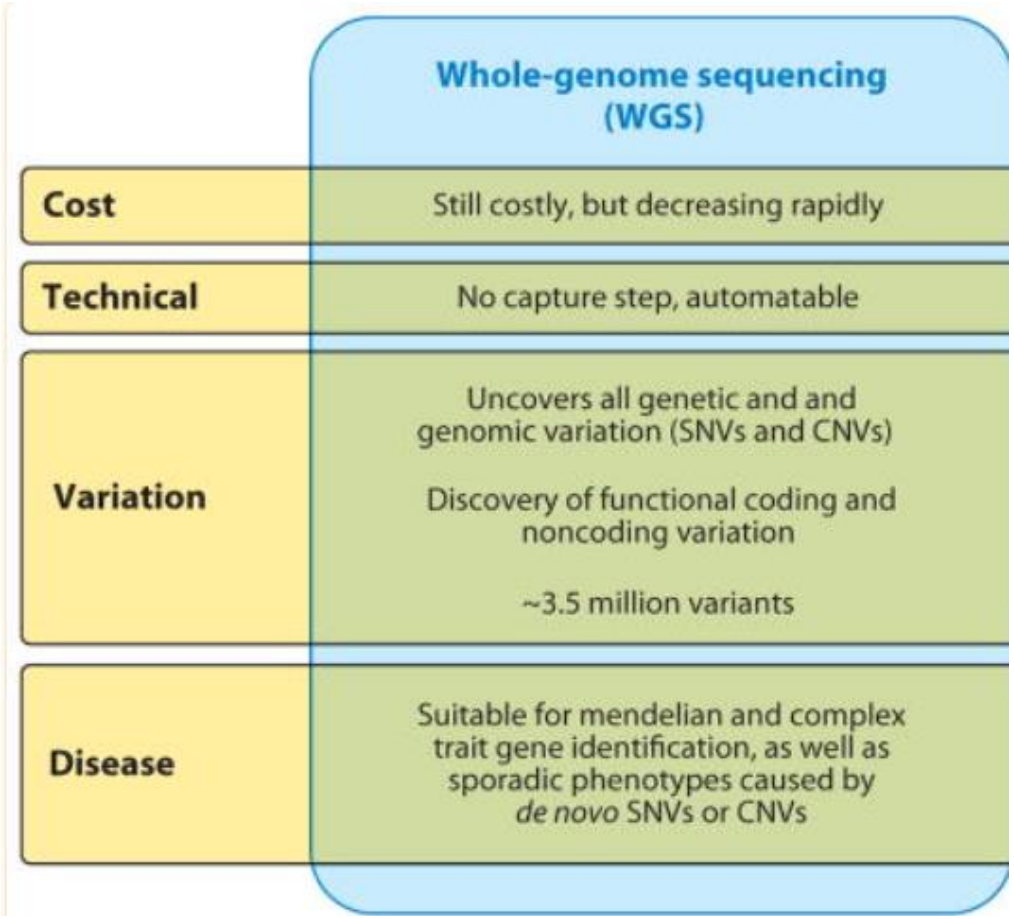
Health Genometer Smart Plan:

- Comprehensive health risk assessment
- Personalized Health Guide
- Tailored to individual medical conditions, pathological risks, and genetic predispositions

Result


Number of Individuals
0
10
20
30
40
50
20-24
25-29
30-34
35-39
40-44
45-49
50-54
55-59
60-64
65-69
70-74
75-79
80-84
85-89
90-94
95-99
Age
Atherosclerosis
Diabetes and Diabetes Risk
Metabolic Syndrome
NAFLD
NASH

Reason for less number in Genometer Plan



	Whole-genome sequencing (WGS)
Cost	Still costly, but decreasing rapidly
Technical	No capture step, automatable
Variation	Uncovers all genetic and and genomic variation (SNVs and CNVs) Discovery of functional coding and noncoding variation ~3.5 million variants
Disease	Suitable for mendelian and complex trait gene identification, as well as sporadic phenotypes caused by *de novo* SNVs or CNVs

Conclusion

- **Summary of Findings:**
  - Efficiency, accuracy, and impact of preventive health diagnostics
  - Benefits and challenges of integrating genomic technologies in healthcare
- **Future Directions:**
  - Continued research and development
  - Training for healthcare professionals

Thanks