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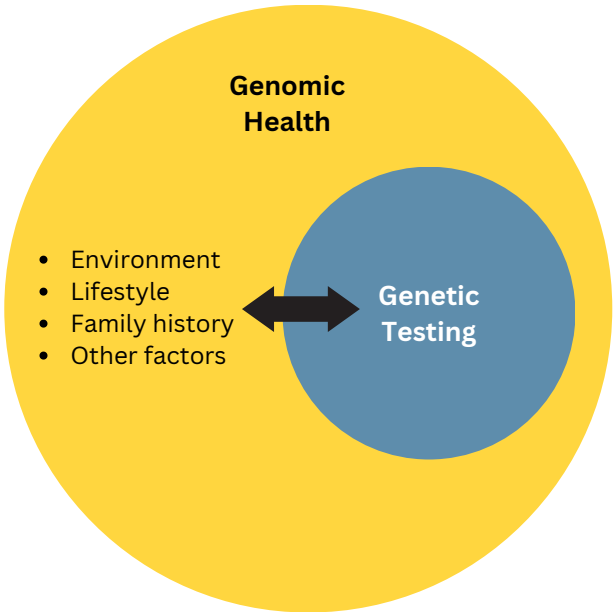
IMPLEMENTATION OF GENOMIC HEALTH IN PRIMARY CARE

1. Genetic testing versus genomic health

Genetic testing refers to the analysis of an individual's DNA to identify specific genetic variations or mutations associated with a particular disease or condition (1).

Genomic health (also known as genomic medicine), on the other hand, is a broader concept that encompasses not only genetic testing but also the analysis of an individual's entire genome to better understand how it influences their overall health and disease risk (2).

Genomic health is an interdisciplinary field that combines genetics, genomics, molecular biology, and bioinformatics to study the interactions between an individual's genes and their environment, lifestyle, family history, and other factors that contribute to their health and disease risk (3).



Written By:
Dr. Yew Sheng Qian

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2. Applications of genomic health

Diagnosis of rare diseases

Genomic health can be used to diagnose patients who have high-risk genetic errors that can cause rare diseases (4, 5).

Diagnosis of common diseases

Genomic health are increasingly being used to understand the genetic factors that lead to the development of common diseases, such as hypertension, diabetes, and cancer (6).

Disease risk assessment

Genomic health can help to identify an individual's risk for developing certain diseases, such as CVD (7) and familial hyperlipidaemia (8), enabling them to take proactive steps to reduce their risk.

Pharmacogenetics

Genomic health may be used to predict whether a person will respond to a particular drug, how well they will respond to that drug, and whether they are likely to get any side effects from the use of a specific drug (9).

Prenatal testing

Prenatal diagnosis of genetic diseases allows parents to make decisions about whether to continue with the pregnancy. It also allows early diagnosis and possible treatment of genetic disease in utero or at birth (10).

Infectious diseases

Sequencing the genomes of microorganisms that cause human infection can identify the exact organism causing the disease, help to trace the cause of infectious outbreaks, and give information as to which antibiotics are most likely to be effective in treatment (11).

Personalised medicine

Personalised medicine describes the use of genetic information to tailor health care intervention to individual need (12).

Gene therapy

Gene therapy involves the administration of DNA or RNA in order to correct a genetic abnormality or modify the expression of genes. Genome editing can add in, cut out, or replace sections of the DNA sequence (13).

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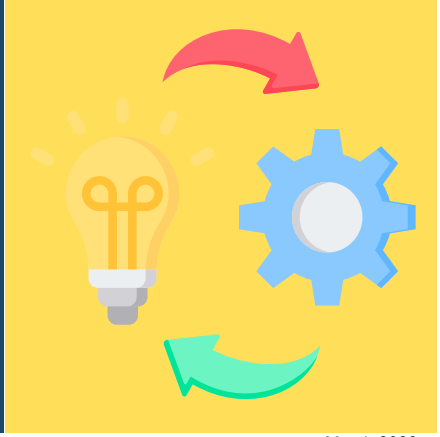

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3. Implementation process of genomic health in primary care

Step 1: Qualitative studies with primary care providers (PCPs)	• Conduct qualitative interviews to explore PCPs' understanding regarding genomic health (e.g., how they cope with new genetic technologies, what are their perceptions of implementing genetic testing, what are their experience in genomic health, etc).
Step 2: Develop and distribute questionnaires to PCPs	• Transcribe the qualitative interviews (in Step 1) and develop a questionnaire to gather PCPs' opinions on the genomic health. In addition, assess their confidence in collecting family history and in providing advice for genetic test results in the primary care setting.
Step 3: Qualitative studies with patients	• Conduct qualitative interviews to identify patients' response and perceptions (i.e., perceived benefits and perceived adverse effect) towards genomic health.
Step 4: Systematic review	• Conduct a systematic review to determine the gap in knowledge in genomic health (e.g., explore which disease is yet to have genetic testing). • Researchers shall also identify the effects (beneficial and adverse effects) of genomic health towards patients' health behaviour. • Using these findings, researchers must convince their funders regarding the need of genomic health in primary care.
Step 5: Develop genomic health intervention tools	• Develop the intervention tools (e.g., genetic testing, risk prediction engines, questionnaire on family history, etc) and apply these tools to patients with familial disease risk.
Step 6: Outcome assessment	• Determine any improvement in identification of patient at risk at primary care clinics. • Measure any improvement in patients' surveillance (i.e., how many patients are referred to specialist clinic for further investigation and management). • Identify the presence or absence of adverse effect (physical and psychological) among the participants.

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4. Challenges of implementing genomic health in primary care setting

- Limited evidence and conflicting interpretation of benefits of genomic health
- Lack of institutional and clinician acceptance
- Lack of standards for genomic applications
- Limited access to genomic health expertise and testing
- Lack of EMR integration of genomic results and clinical decision support
- Poor documentation of family history in primary care clinics

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BIG DATA IN HEALTH CARE: WHAT IT IS?

By Dr Nur Aazifah bt Ilham

Big data is a term often used to describe an explosion of information. In order to be classified as big data, a dataset should fulfill the following criteria as below:

High Volume (Scale of data)

Usually in terabytes or petabytes which are managed and stored using Hadoop or Apache Spart.

High Variety

(Different form of data)

The format of data can be structured or unstructured.

High Velocity

The data are frequently produced and analyzed.

Veracity (Uncertainty of data)

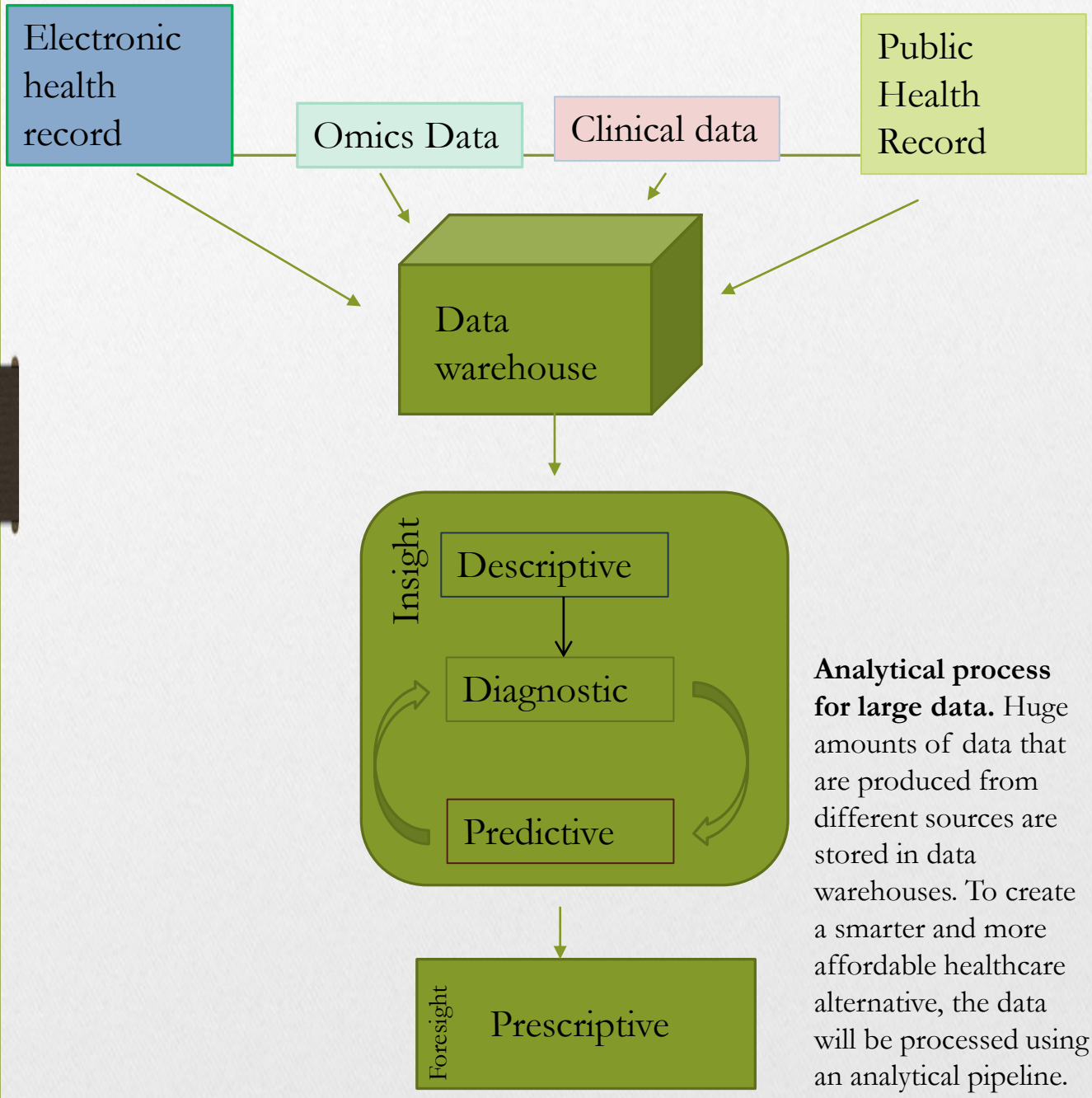
Uncertainty due to data inconsistency, incompleteness, and latency. Thus, the quality, relevancy, predictive value and meaning of data might be questioned.

Value

The information is valuable to various stakeholders or decision makers.

Healthcare as big data repository

Healthcare is multidimensional and complicated system with the objective of prevention, diagnosis, treatment and rehabilitation of disease. The massive date from various stakeholders, including the health professionals, health facilities, and financing institutions is valuable in helping decision making and improving health outcome.



Analytical process for large data. Huge amounts of data that are produced from different sources are stored in data warehouses. To create a smarter and more affordable healthcare alternative, the data will be processed using an analytical pipeline.

Smarter and cost-effective decisions will improve outcome

Adapted from Dash et al (2019) Big Data in Healthcare: Management, analysis and future prospects J Big Data

Big data application in healthcare

Administration and healthcare delivery

- Big data analytic can be used for management of healthcare to improve efficiency in delivery of service and cost-effectiveness.
- Example: Prediction of the requirement number of staff required based on previous past information can reduce patient waiting time.
- Cost-effectiveness analysis from big data will aid policy makers to make decisions to improve health outcome with reduced cost.

Clinical Information & Clinical decision support

- Health information from structure and unstructured data will be merged to help develop clinical decision. This will increase the accuracy of diagnosis, hence, improving the management plan.
- This can be done using a well plan diagnostic and predictive study. This may ease translational practice to occur in the local study.

Integrating big data with medical imaging

- Machine learning has been used to help to diagnose disease from billions of images. However, machine learning technique requires a huge number of images to accurately learn to make the diagnosis.

Personalized/precision treatment

- Systematic and integrative analyses of omics data in conjunction with EMR integration can help to design a better treatment towards personalized/precisian medicine.

Internet of Thing (IoT) devise

- IoT devise create a continuous stream of data which can be used as a health monitoring. Such devise is beneficial to be used by elderly and patients with chronic illnesses.
- The patient's parameter can be integrated in the EMR which can be used to predict health status.

Challenges in big data analysis

Big data generates distinct features that do not present in traditional datasets. Below are the major challenges of big data:

The complexity of big data give a unique statistical impact and computing infrastructure. However, if the difficulty can be overcome, the advantages of using big data are huge and unimaginable.

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