

#### Singapore launches next phase of National Precision Medicine Strategy

- 1. Precision Medicine is a data-driven medical approach which takes individual variations in genetics, environmental and lifestyle factors into account, allowing doctors to more accurately predict which treatment and prevention strategies will work in different groups of people.
- 2. Enabled by tools to analyse data on a large scale and with DNA sequencing becoming more affordable, Precision Medicine can improve healthcare by taking the guesswork out of disease treatment, minimising side effects, reducing wastage of resources to deliver targeted treatments for better outcomes, thus improving public health which everyone can benefit from.
- 3. Precision medicine is a key focus of the Human Health and Potential Domain under the Singapore's Research, Innovation and Enterprise (RIE) 2025.
- 4. **Precision Health Research, Singapore (PRECISE)** is the central entity set up to coordinate a whole of government effort to implement Phase II of Singapore's 10-year National Precision Medicine (NPM) strategy.
- 5. This is a concerted effort at the national level to build on the foundations of Phase I of the NPM, where work began in 2017 to sequence 10,000 Singaporean genomes in what was known as the **SG10K** population health study to address the lack of Asian genomic data. Today the **SG10K** Health Study is the largest multi-ethnic Asian database to-date.
- 6. Phase II of the NPM commenced in 2020, and PRECISE has partnered Professor John Chambers at Nanyang Technological University's Lee Kong Chian School of Medicine in this landmark population health research study to sequence the genomes of 100 000 healthy Singaporeans (SG100K) and 50,000 people with specific diseases. Phase II will also include pilot clinical implementation of precision medicine approaches and establish data infrastructure for the linkage of genomic data with electronic health records and other data types.
- 7. In addition, NPM Phase II will also enhance the breadth and depth of the Precision Medicine-related industry by attracting and anchoring overseas companies in Singapore, while yielding new opportunities for home-grown companies.



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#### 8. Singapore's NPM Strategy in a nutshell:



- 9. Benefits of Precision Medicine:
  - ACCURATE PREDICTION: Reliable biomarkers help doctors pre-empt possible health conditions and act before it is too late
  - **FASTER DIAGNOSES:** De-mystify the causes of rare diseases and accelerate the search for potential treatments
  - **OPTIMISED TREATMENTS:** Patients receive the right drug at the right time, improving efficacy and reducing side-effects
  - **NEW TREATMENTS:** Understanding the basis of disease will inform the development of new drugs and therapies



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10. Researchers, company representatives and citizens who are keen to participate in the SG100K population health study, please contact PRECISE at <u>contact@precise.cris.sg</u>.

For more information, visit https://www.npm.sg. (Refer to Annex B for Case Study on how the use of Precision Medicine as a medical approach has been useful in the discovery and treatment of rare childhood diseases)



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#### Annex A

Photo 1 of 2:



Established to coordinate a whole of government effort, Precision Health Research, Singapore (PRECISE) seeks to improve patient outcomes, implement data-driven healthcare and capture economic value through precision medicine. (Photo credits: PRECISE)

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Photo 2 of 2:



Precision medicine takes individual variations in genetics, environmental and lifestyle factors into account, allowing doctors to more accurately predict which treatment and prevention strategies will work in different groups of people. (Photo credits: PRECISE)

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#### Annex B

Precision medicine has seen success in the treatment of rare genetic diseases in Singapore. For over two decades, the Genetics Service at KK Women's and Children's Hospital (KKH) has been providing clinical care to paediatric patients with genetic disorders, tapping on a precision medicine approach to disease treatments.

Through BRIDGES (<u>B</u>ringing <u>R</u>esearch <u>I</u>nnovations for the <u>D</u>iagnosis of <u>GE</u>netic diseases in <u>S</u>ingapore), 412 families were analysed over the last six years by genomic researchers at SingHealth, A\*STAR and Duke-NUS. As a result, 160 children with rare and undiagnosed conditions were able to receive proper diagnoses that led to tailored clinical management and improved health outcomes in at least one of three families.

Precision medicine has helped to improve understanding of the causes to genetic disorders, and helped clinicians to assess and identify evidence-based, therapeutic targets for developing precise treatments to the conditions.

#### **Case Studies:**

From 2014 to 2020, BRIDGES (<u>Bringing Research Innovations</u> for the <u>D</u>iagnosis of <u>GE</u>netic diseases in <u>S</u>ingapore) through the use of precision medicine approach has helped 160 children with rare and undiagnosed conditions to receive proper diagnoses, and enabled doctors to provide better care to them and their families.

Also, six of these children were diagnosed with a new disease that had never been reported before, with two of these discoveries being led by clinicians at KK Women's and Children's Hospital (KKH) and scientists at A\*STAR:

- Jamuar Syndrome the world's first known case of a genetic syndrome in two Indian sisters with a unique clinical profile that presented epileptic seizures and developmental delays. Jamuar Syndrome was named after Dr Saumya Jamuar, who first recognised the clinical features as a new disorder. Dr Jamuar is Senior Consultant, Genetics Services, Department of Paediatrics at KKH and Head of SingHealth Duke-NUS Genomic Medicine Center.
- 2. *PKB Syndrome* the first human report of mutation in the *EIF6* gene in a six-year-old Chinese boy who had features like those for Shwachman-Diamond syndrome (SDS), a rare condition characterised by pancreatic insufficiency and bone marrow failure. The clinical symptoms had not



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previously been associated with the *EIF6* gene and the PKB Syndrome was named after Professor **P**hua **K**ong **B**oo, who first identified the unique cluster of symptoms. Professor Phua is Emeritus Consultant, Gastroenterology, Hepatology & Nutrition Service, Department of Paediatrics at KKH.



Instituting precision medicine approach in clinical pathways, Dr Saumya Jamuar analysing a patient's genomic data. (Photo credits: KK Women's and Children's Hospital)

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