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Precision Medicine Brings Hope for Rare Disease Care

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Precision medicine is redefining rare disease care across Asia-Pacific. By combining genomics, artificial intelligence and regional collaboration, healthcare systems are moving towards earlier diagnosis, more accurate treatment and stronger clinical capabilities, helping address one of the region's most significant unmet healthcare challenges.



For patients living with a rare disorder, receiving an accurate diagnosis can take years. Misdiagnosis often leads to unnecessary treatments, avoidable side effects, and a severely diminished quality of life. Of the more than 7,000 known rare diseases, only 5 per cent have an approved therapy. Fortunately, precision medicine is rapidly closing the gap between clinical need and reality.

Across Southeast Asia alone, an estimated 45 million people live with a rare condition, and over 10 million remain undiagnosed. Compounded by ageing populations and persistent infrastructure gaps, traditional healthcare models across the Asia-Pacific region cannot keep pace. This reactive approach forces patients and their families into a prolonged "diagnostic

odyssey" that drains finances, tests emotional resilience, and often ends without answers.

Driven by dynamic alliances spanning patient advocacy groups, NGOs, and public-private partnerships, APEC economies are moving toward a proactive, healthcare model. It anchors in early genetic screening that can detect rare conditions before symptoms appear. Because these conditions are individually rare but collectively common, they demand a coordinated, cross-border response.

One promising framework is the HGP2 Rare Disease Alliance (RaDiAnce-APAC), which unites health authorities across 11 countries. By accelerating the clinical translation of advanced genomic technologies, the alliance aims to systematically close the region's diagnostic gap, transforming rare disease management from isolated efforts into a unified regional strategy.

In Indonesia, a collaboration with Dr. Cipto Mangunkusumo National General Hospital (RSCM) applied Whole Exome Sequencing (WES) to decode DNA and pinpoint disease-causing mutations in paediatric rare diseases. Subsequent programmes with RSAB Harapan Kita Women and Children Hospital are introducing Copy Number Variation Sequencing (CNV-seq), enabling clinicians to detect complex chromosomal abnormalities long before symptoms emerge. This provides families with a critical head start on intervention.

For precision medicine to achieve a true public health impact, screening must evolve from localised pilots into national policy. Thailand offers a compelling case study. Non-Invasive Prenatal Testing (NIPT), which flags severe fetal conditions through a simple maternal blood draw, has been integrated into the country's universal healthcare system. Following the launch of the Southeast Asia Thalassemia Prevention and Control Alliance, Thailand's Ministry of Public Health also enacted dedicated Advanced Therapy Medicinal Products (ATMPs) policies, creating comprehensive screening and treatment frameworks for genetic disorders.

Rare diseases are notoriously difficult to catalog, often requiring geneticists to comb through millions of data points to find a single pathogenic variant. This is where artificial intelligence (AI) is transforming the field. AI can now analyse massive datasets simultaneously, identifying complex clinical signatures that human curators might overlook. BGI Genomics' GeneT AI has advanced to clinical implementation specifically to address this challenge. By optimising pathogenic variant selection, these tools substantially reduce the cost of genetic data interpretation.

GeneT empowers variant curators with an end-to-end workflow, from initial interpretation to automated reporting. Its multi-level bioinformatics analysis covers single nucleotide variants (SNVs), copy number variations (CNVs), and mitochondrial mutations, enabling a seamless transition from raw genomic data to the final clinical report.

Building long-term capabilities and resilience requires sustained talent cultivation. Initiatives like the "International Genetic Disease Interpretation Training Programme" are addressing this gap, bringing together medical personnel from Thailand, Indonesia, and Vietnam to deepen the local understanding of complex genetic pathways. Through joint laboratories, technology transfer, and localised education, regional partners are transforming innovation into self-reliant, lasting public health capacity.

As APEC charts the region's economic and technological future, precision medicine into rare disease care stands as a powerful testament to collaborative innovation. By committing to shared experiences, AI-driven diagnostics, and local capability building, the Asia-Pacific community can finally end the diagnostic odyssey for millions of underserved patients.

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