養和醫療
HKSH MEDICAL GROUP

For Immediate Release

HKSH Offers In-House "Comprehensive Cancer Genomic Testing"
Pairing Strengths in Molecular Genetics and Cytogenetics for Cancer Management

(20 May 2025, Hong Kong) Precision cancer treatment is based on accurate pathological diagnosis and cancer type classification. Harnessing advanced technologies and professional expertise at both genetic and chromosomal levels, The HKSH Cancer Centre (HKSH) is currently the only private medical institution in Hong Kong that offers comprehensive cancer genomic testing. By integrating *Comprehensive Genomic Profiling (CGP)*, which identifies gene mutations, and *Cytogenetics*, which reveals chromosomal abnormalities, into clinical services, this dual approach offers critical insights into tumour pathogenesis. From diagnosis to treatment and even in cases of relapse, the use of CGP and cytogenetics not only enhances diagnostic accuracy and prognosis evaluation, but also allows doctors to tailor therapies, monitor treatment efficacy and drug resistance, achieving personalised precision medicine through the decoding of genes and chromosomes.

Two-Tier Cancer Genomic Testing

Tier 1: Cancer Specific Panels for Five Cancer Types

To address cancer-related genetic mutations, HKSH has been utilising Next Generation Sequencing (NGS) technology since 2013. HKSH developed Cancer Specific Panels for five cancer types, including blood cancer, lung cancer, brain cancer, colorectal cancer, and endometrial cancer. This targeted approach focuses on identifying genetic mutations specifically associated with each cancer type, offering the advantages of short turnaround time, high accuracy which helps accelerate diagnosis and guide more informed treatment decisions.

Taking lung cancer as an example, HKSH performed over 930 tests for this cancer between August 2023 and December 2024. Among this cohort, 54.6% revealed EGFR (Epidermal Growth Factor Receptor) mutation, a common genetic mutation in lung cancer, indicating that targeted therapy could be a possible treatment option. With the increasing availability of targeted therapy and immunotherapy options, Cancer Specific Panel testing allows for early detection of actionable mutations, enabling doctors to formulate precision treatment strategies, manage potential drug resistance, and ultimately improve patient outcome and their quality of life.

Tier 2: Comprehensive Genomic Profiling (CGP)

To further enhance the efficiency of cancer genomic testing and fully realise the capabilities of NGS, HKSH's Department of Pathology launched the CGP service in November 2024. HKSH is currently the only private hospital in Hong Kong capable of conducting the entire testing process in-house from tissue sampling, laboratory testing to data analysis, without the need to send samples to overseas laboratories. In-house CGP facilitates timely and in-depth genomic analysis for patients with advanced cancers or complex clinical conditions, enabling more personalised and precise treatment strategies.

Dr. Edmond MA, Director of Clinical Pathology & Molecular Pathology Division and Specialist in Haematology, Hong Kong Sanatorium & Hospital, explained, "CGP plays a crucial role in three key areas for advanced, relapsed refractory, and metastatic cancers. First, it helps identify actionable mutations for matching targeted therapy. When specific mutations are detected in a tumour's genetic profile, doctors can tailor personalised treatment plans accordingly. Second, CGP provides valuable insights into a patient's prognosis. Certain genetic biomarkers can help predict how a tumour is likely to respond to specific treatments. For instance, if CGP suggests a tumour is unlikely to respond to chemotherapy, doctors can then explore alternative treatment options early on, sparing patients from unnecessary side effects. However, the genetic characteristics of tumours are not completely static, such as in cases of drug resistance. In non-responders or relapsed patients, cancer genetic testing can reveal genetic changes associated with drug resistance, helping doctors to adjust treatment strategies timely and keep the condition under control."

HKSH's CGP service offers broad genomic coverage in a single test, short turnaround time and user-friendly reports with interpretative comments. CGP enables the detection of mutations in 335 cancer-related genes in one single assay, uncovering critical biomarkers and tumour characteristics such as Microsatellite Instability (MSI), Tumour Mutational Burden (TMB), and Homologous Recombination Deficiency (HRD). This comprehensive approach requires only a small tissue sample, minimising the need for patients to undergo multiple biopsies.

Samples of cancer genetic testing are generally sent to overseas laboratories. HKSH could now perform the entire testing process in-house, thus reducing the reporting time. The testing technology used by HKSH is also aligned with overseas CGP technologies with connectivity to the same database, ensuring equally high reference value. Furthermore, instead of just listing genetic abnormalities, HKSH's CGP reports integrates each patient's clinical context with expert pathological analysis to deliver reports with a clear, succinct, and personalised narrative, offering specific guidance for precision medicine.

Dr. YAU Chun Chung, Director of Department of Radiotherapy, Associate Director of Comprehensive Oncology Centre and Specialist in Clinical Oncology, Hong Kong Sanatorium & Hospital, has managed a number of cancer cases by using CGP. He said, "Patients with advanced or recurrent cancer need a clear treatment direction. HKSH is now equipped with in-house CGP capabilities which enable us to understand the characteristics of genetic mutation profiles of the cancer cells so as to match patients with the most appropriate therapies. The shorter turnaround time is also a major benefit for patients." *(Patient cases on next page)*

Integrating Chromosomal Analysis: Dual Decoding of Cancer Genomics

The human body is composed of trillions of cells. Within each cell nucleus, the DNA is packaged into 46 chromosomes, each carrying important information on heredity. Today, comprehensive cancer genomic testing allows a deeper, more accurate understanding of the disease by analysing the genes and chromosomes of cancer cells. This approach helps uncover vital insights into genetic mutations, chromosomal abnormalities (such as deletions, duplications, and translocations), and tumour evolution.

In addition to gene mutations, chromosomal abnormalities offer critical insights into the biology of cancer. Cytogenetic analysis, the study of chromosomes, is especially important in diagnosing and managing haematological malignancies. By examining individual cells, cytogenetics enables rapid and accurate, genome-wide testing, identifying chromosomal translocations, copy number variations, and amplifications that may drive cancer development. **Dr. Thomas WAN, Molecular Pathology Division Supervisor and Cytogeneticist of Hong Kong Sanatorium & Hospital**, elaborates: "One key method is karyotyping, which involves visualising the size, shape, and number of chromosomes under a microscope. Cancer cells often display complex chromosomal patterns. An example is the Philadelphia chromosome, the cause of chronic myeloid leukaemia, which arises from a translocation between chromosomes 9 and 22, resulting in the formation of the BCR::ABL fusion gene." Another widely used cytogenetic technique is Fluorescence *In-Situ* Hybridisation (FISH). This method employs fluorescent probes to identify the presence, absence, or structural changes of specific DNA sequences within chromosomes.

Leveraging Artificial Intelligence (AI) to Boost Diagnostic Efficiency

Traditionally, karyotyping involved culturing cells, followed by labour-intensive manual analysis under an optical microscope. Medical technologists would visually inspect, trim, and manually arrange chromosomes to create a karyotype map, which is a process that required substantial time and manpower, ultimately delaying results. The integration of automation and AI has revolutionised this workflow. Through training and learning, advanced software can now automatically identify and classify metaphase chromosomes, capture digital images and generate karyotype maps. These outputs are then verified by medical professionals, significantly enhancing the efficiency of the diagnostic process.

While CGP focuses specifically on analysing gene mutations, Cytogenetic analysis, which examines the entire tumour genome through chromosomal profiling, complements CGP to provide a more holistic understanding of the mechanisms underlying oncogenesis beyond gene mutations. HKSH's Department of Pathology integrates these two cutting-edge in-house cancer diagnostic technologies which complement each other to deliver critical insights for doctors and patients, enabling targeted and effective cancer treatment.

Dr. Joseph CHAN, Chief Medical Officer of HKSH Medical Group and Deputy Medical Superintendent of Hong Kong Sanatorium & Hospital, remarked, "Comprehensive cancer genomic testing is a crucial first step in effective cancer management. The HKSH Cancer Centre is equipped with cutting-edge diagnostic and treatment technologies, allowing doctors to formulate precise and effective treatment plans based on genetic testing results and each patient's condition. Whether patients require surgery, chemotherapy, proton therapy, stereotactic radiotherapy, targeted therapy, hormonal therapy, cellular therapy, or immunotherapy, our dedicated medical team stand ready to provide comprehensive and compassionate support throughout the treatment and recovery journey."

Patient Case 1

Mr. Cheung, 63 years old, experienced a relapse of non-small cell lung cancer (NSCLC) in 2023. Genetic testing revealed a ROS1 gene fusion, allowing him to be successfully matched with an oral targeted therapy that effectively controlled the disease. However, in early 2025, rising tumour markers and the onset of chest pain raised concerns. A PET-CT scan confirmed a relapse. To investigate further, a CT-guided biopsy was performed, followed by CGP. The analysis identified cancer cell originally carrying CD74-ROS1 fusion gene developed a G2032R mutation, which had rendered the original targeted therapy ineffective due to drug resistance. With these genomic insights, Mr. Cheung's treatment was promptly adjusted. He was prescribed another targeted therapy specifically designed to tackle G2032R mutation and his condition has improved significantly. His chest pain has resolved, and follow-up X-ray imaging shows a substantial reduction in tumour shadows.

Patient Case 2

Ms. Ho, over 90 years old, was first diagnosed with urothelial carcinoma in 2020 and achieved disease control following initial treatment. Unfortunately, the cancer relapsed in 2025, having metastasised to nearby lymph nodes, which led to swelling, pain, and ulceration. Due to her advanced age, Ms Ho's body could not bear the side effects of chemotherapy so her medical team opted for a lymph node biopsy, followed by CGP. The analysis revealed a BRCA1 gene mutation, commonly associated with hereditary breast cancer, along with evidence of homologous recombination deficiency (HRD). Based on these findings, Ms. Ho was prescribed PARP inhibitors, a targeted therapy designed specifically for cancers with BRCA mutations. The results have been remarkable with significant tumour reduction and healing of the ulcerated area. Thanks to the therapy's targeted mechanism of action, side effects have been minimal, enabling Ms. Ho to maintain a reasonable quality of life.

–END–

For media enquiries, please contact
Corporate Affairs Department

Carol KWOK Tel: 2917 5838
Joyce CHAN Tel: 2917 5835
Paley WONG Tel: 2917 5837

Email: media@cad.hksh.com

養和醫療
HKSH MEDICAL GROUP

About HKSH Medical Group

Officially launched in September 2017, HKSH Medical Group, through its operating members, adopts a multi-faceted, coordinated approach to promote public health and advanced medicine through relentless efforts in clinical excellence, medical training and research as well as public health education. Members of the Group, including Hong Kong Sanatorium & Hospital, HKSH Healthcare, HKSH Eastern Medical Centre and HKSH Cancer Centre, are dedicated to offering top-quality holistic care to patients, upholding the motto 'Quality in Service, Excellence in Care.'

Established in 1922, Hong Kong Sanatorium & Hospital is one of the key members of HKSH Medical Group and a leading private hospital in Hong Kong. Living up to its motto of 'Quality in Service, Excellence in Care', the Hospital is committed to serving the public as well as promoting medical education and research.

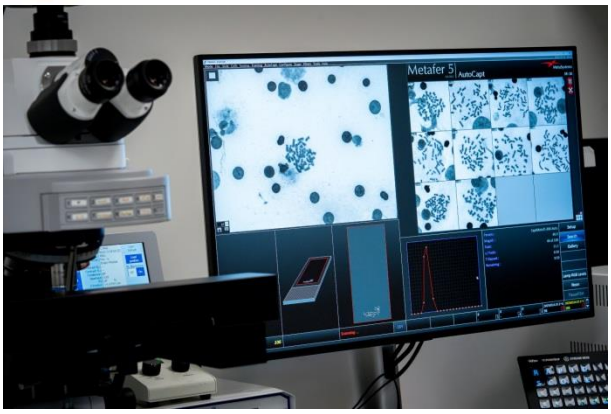
For more information about HKSH Medical Group, please visit http://www.hksh.com.

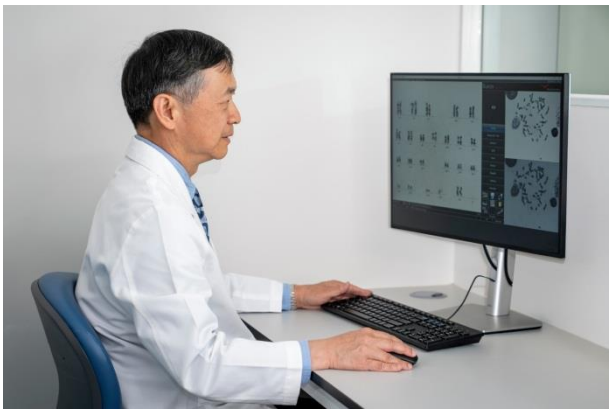
Photo Captions:

1. **Dr. Joseph CHAN, Chief Medical Officer of HKSH Medical Group and Deputy Medical Superintendent of Hong Kong Sanatorium & Hospital** (second from right), **Dr. Edmond MA, Director of Clinical Pathology & Molecular Pathology Division and Specialist in Haematology, Hong Kong Sanatorium & Hospital** (second from left), **Dr. Thomas WAN, Molecular Pathology Division Supervisor and Cytogeneticist of Hong Kong Sanatorium & Hospital** (first from left), **and Dr. YAU Chun Chung, Director of Department of Radiotherapy, Associate Director of Comprehensive Oncology Centre and Specialist in Clinical Oncology, Hong Kong Sanatorium & Hospital** (first from right), stated that HKSH has incorporated *Comprehensive Genomic Profiling* and *Cytogenetics* into clinical services. This dual approach helps decode both genetic mutations and chromosomal abnormalities in cancer cells, enabling precise and personalised treatment strategies.

2. HKSH is currently the only private hospital in Hong Kong that offers in-house Comprehensive Genomic Profiling. Compared to sending tissue samples to overseas laboratories, in-house testing shortens turnaround time, optimises the use of tissue samples, and enhances cost-effectiveness. The pathology team can also provide reports with a clear, succinct, and personalised narrative, offering specific guidance for precision medicine.

3. Dr. Edmond MA stated that HKSH's Department of Pathology is currently equipped with four Next Generation Sequencing (NGS) machines and HKSH utilises NGS in both Cancer Specific Panels and Comprehensive Genomic Profiling (CGP), which shortens the required time compared to sending tissue samples to overseas laboratories for CGP testing.





4. HKSH has integrated AI into cytogenetics to assist in identifying metaphase cells and performing karyotype analysis, which has significantly reduced manual workload and turnaround time, enabling the delivery of detailed reports within just 3 to 7 days.