Haematology opens Next Generation Sequencing Facility

The Department of Haematology, Christian Medical College, Vellore, and MedGenome, genomics based research and diagnostics company, have come together to collaborate on research projects using the Next Generation Sequencing (NGS) platform.

The Department of Haematology at CMC, Vellore has been at the forefront of research in blood related disorders.

With a recently sanctioned 'Centre of Excellence in Haematology' Program by the Department of Biotechnology, New Delhi, India, this NGS facility is getting established.

With their expertise in sequencing and research background on the NGS platform, MedGenome will support the department in all aspects of NGS analysis including the Bioinformatics pipeline and interpretation of results.

This collaboration will help CMC Vellore, not only to gain rich genomic insights but also enable better understanding of the disease at molecular level and translation of insights into clinical practice.
The following are the major projects that will be carried out using this NGS facility:

Potential diagnosis and research applications of NGS in the non-malignant haematological conditions

- Exome sequencing to identify rare and novel genes involved in the pathogenesis of Fanconi Anaemia
- RNAi screen by NGS to study the mechanisms of human erythropoiesis
- Chip-seq to study transcription factors in human erythropoiesis

- Exome sequencing/ targeted resequencing to identify mutations in patients with Primary immunodeficiencies (PIDs) to facilitate cost effective and earlier diagnosis where a definitive diagnosis is difficult due to overlapping symptoms
- Screening of inherited genetic defects in aplastic anemia in children
- Iron related disorders can be due to genetic defects in genes involved in iron metabolism. A comprehensive panel will help to identify these genetic defects

<u>Potential diagnosis and research applications of NGS in the area of leukemia / HSCT</u>

- A diagnostic panel for commonly occurring and clinically relevant mutations in Acute Myeloid Leukemia (AML) in a prospective cohort of patients with AML diagnosed and treated at our centre with uniform protocol
- Exome sequencing in imatinib intolerant or resistant CML to identify a genetic signature predicting imatinib intolerance / resistance
- NGS based screening of BCR-ABL tyrosine kinase mutations in TKI resistant CML patients
- Screening of newer molecular markers in ALL for further prognostication/ risk stratification
- Targeted resequencing of drug metabolism associated genes to identify the genetic variants predicting conditioning regimen related toxicity in patients undergoing HSCT.

Read news reports of the facility in The Hindu and The New Indian Express.