

## **First unit to examine DNA for mutations in genes set up at SCTIMST, Kerala**

Sree Chitra Tirunal Institute for Medical Sciences and Technology (SCTIMST) has established a Molecular Genetics and Neuro-immunology Unit (MGNU) for genetic testing of selected cardiac and neurological inherited diseases.

This unit which will examine DNA for mutations in genes that may cause specific diseases has been set up with support from Department of Science and Technology (DST). The unit will start functioning from February 26, 2020.



SCTIMST is an advanced referral centre under DST, for neurological and cardiovascular diseases and patients are referred to the centre not only from all over Kerala but also other neighbouring states for the management of such genetic diseases.

This diagnostic platform will be available to patients from SCTIMST and hospitals outside specifically for multi-gene panels for diseases such as Neuromuscular diseases, Movement disorders (Parkinson's Disease, Huntington's Disease), Neuro-developmental disorders, Epilepsy syndromes, Inherited metabolic diseases, Mitochondrial diseases and for identification of bacteria from body fluids.

Some genetic diseases run in the family, and others occur 'de novo' (start with the person who is affected). Besides diagnosis, advanced genetic testing also plays a vital role in determining the risk of developing certain diseases, screening of diseases and for guidance in instituting appropriate medical treatment. Testing the inheritance of diseases in a family can help the family in planning for the future and also encourage other members in the family to get tested. However, testing of unaffected family members will be offered only after proper counselling if necessary.

With the establishment of these advanced high-throughput sequencing technologies, it has become possible to sequence more genes and more samples in a single run. This will overcome the limitations in terms of costs, speed and scale, which will benefit clinicians and patients alike.

This diagnostic facility opens new avenues for extensive research and genomics and aids existing and new research projects, including the conduct of collaborative research work within and outside the country. The unit has been equipped with the most advanced facilities to perform the genetic diagnostic tests using Real-Time Polymerase Chain Reaction, Sanger sequencing and Next-Generation sequencing. This is the first centre with such an advanced sequencing platform in Kerala.

The long-term plan of the genetic diagnostic unit is to establish a robust bioinformatics platform to provide support for disease diagnosis and management, molecular characterization of diseases for personalised treatment, prenatal screening for fetal genomic disorders, population screening for disease risk, pharmacogenomics (study of how genes affect a person's response to drugs), identification of novel genetic biomarkers and genetic counselling.