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(एक राष्ट्रीय महत्व का संस्थान, विज्ञान एवं प्रौद्योगिकी विभाग, भारत सरकार)
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P&A.III/LOK SABHA/211/SCTIMST/2021

21.07.2021

Shri. Jatin Singh
ASO
DHR
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Sir,

Sub :- Lok Sabha Admitted Starred QUESTION No. 70 (Dy. No. 1563) - reg.
Ref :- E-mail dtd. 19.07.2021 from Manoranjan Mohanty , Director/Scientist
F, AI Division

Please find below the information relating to SCTIMST on the subject referred above:

Question (a) Whether the Government proposes to ramp up dedicated research & development on rare genetic diseases like spinal muscular atrophy and others along with the deployment of available resources for treatments to get maximum health gains for patients suffering from such diseases;

(b) If so, the details thereof ;

Answer to a &b:

1. The proposal for starting a genetic neuromuscular registry was submitted. This was approved by Technical Advisory Committee and is awaiting clearance from Institute Ethics Committee. This registry includes conditions like spinal muscular atrophy, muscular atrophies etc. and helps in streamlining multidisciplinary care.

2. A research proposal for determining the frequency of Pompe disease among undiagnosed muscle diseases is submitted to Institute Ethics Committee.
3. An intramural study on genotype - phenotype correlation in Charcot Marie Tooth disease (genetic neuropathies) is IEC approved and ongoing.
4. The Department is exploring the possibilities of obtaining costly medicines for spinal muscular atrophy under compassionate use programmes.
5. Pediatric neurology subsection of Department of Neurology is doing a research on 'clinical and radiological profile of genetic leukencephalopathies in children and adults'. These are rare neurogenetic disorder causing white matter changes in brain.
6. GENETICS OF COMPLEX PEDIATRIC EPILEPSY SYNDROMES: ELECTRO CLINICO-IMAGING BASED GENOTYPE- PHENOTYPE CORRELATIONS IN AN INDIAN COHORT- Funded by ICMR- Allocated cost INR 1.5 crore. This study aims to analyze the genetic basis of a wide spectrum of complex epilepsies with age of onset spanning from the neonatal period to late childhood (<12 years age). We plan to study a cohort of cases with varied phenotypes classified based on clinical evaluation including developmental and cognitive assessments, video-EEG and neuroimaging from children with specific epilepsy syndromes. We are collecting probands' and their parents' blood samples presenting with different phenotypes that fall under the rubric of EE or unexplained refractory epilepsy. Demographic details, family history, detailed clinical history are recorded and where appropriate, results of metabolic evaluation if done as part of the standard of care in these children will be collected. Whole-exome-sequencing-based deep sequencing of epilepsy panel genes of trios will be done. Endophenotyping using clinical evaluation, results of metabolic evaluation, EEG and multimodality neuroimaging (MRI) will be conducted. Therefore, our approach on genetic

basis of the disease will help in resolving the phenotype heterogeneity. Study objectives include:

- a. Streamlining diagnosis by correlating phenotype to genotype .
- b. Streamlining therapeutic intervention-prognostication by correlating phenotype to pharmacogenotype.
- c. Ascertainment of the frequency, type, inheritance patterns of pathogenic or likely pathogenic variants in complex epilepsies.

This work is undertaken by R Madhavan Nayar Centre for Comprehensive Epilepsy Care, SCTIMST in collaboration with Rajiv Gandhi Centre for Biotechnology (RGCB), Thiruvananthapuram

7. Facilities for sequencing the DNA and related research for rare inherited disorders can be done in Molecular Genetics Unit upon request from Clinicians.

Question C: the details of the steps taken by the Government to improve Telehealth services and other Digital Health Services for citizens across the nation in order to achieve " Health for all"

Answer to C:

1. Yes, the central government has initiated the National Digital Health Mission (NDHM) to create a cloud-based electronic Health ID for every citizen. In the first phase, it launched in all the Union Territories on August 15, 2020. This is the first step toward creating a standardized digital health infrastructure in the country. <https://ndhm.gov.in/>. The online registry for all healthcare professionals and health facilities is also planned in this initiative.
2. The government has launched the e-Sanjeevini program to leverage telemedicine facilities to expand healthcare coverage. The release of the

Telemedicine Practice Guidelines on March 25, 2020, has brought legal legitimacy to telemedicine consultations in India.

3. The Sree Chitra Tirunal Institute for Medical Sciences and Technology, Trivandrum (an autonomous institute under DST, GoI), has launched an innovative mobile telemedicine program in Wayanad, the sole aspirational district in Kerala, with a substantial proportion of the tribal population. Two mobile telehealth units with a dedicated medical team regularly visit
4. The peripheral health centers and specialist consultation services using the telemedicine facility.
5. The state health department system is dealing with the vaccination program. The Kerala state has started an intensive campaign to catch up with the routine vaccination program from the recent setbacks of the Covid-19 pandemic.
6. The Institute has telemedicine facility since 2005. The National Network Knowledge [NKN] connectivity has been given to the three wings of the Institute to enhance the interaction between clinicians in Hospital wing and Scientists/ Engineers in Biomedical Technology Wing through video conferencing facility. Through NKN facility, Special Continuing Medical Education [CME] program broadcasting with ISRO Telemedicine Network etc is going on.

In order to reduce the risk of COVID-19, by contact with healthcare facilities, other patients, and healthcare staff SCTIMST has started Telemedicine using telecommunications technology and information technologies to provide remote clinical services to its patients since April 2020. All a patient need is a phone or device with the internet to continue the medical care while protecting themselves and the healthcare provider from COVID-19. The use of secure video and audio connections made it

