GOVERNMENT OF INDIA
MINISTRY OF HEALTH AND FAMILY WELFARE
DEPARTMENT OF HEALTH RESEARCH

RAJYA SABHA
UNSTARRED QUESTION NO. 1856
TO BE ANSWERED ON 4TH AUGUST, 2026

RECENT ADVANCES IN GENETIC RESEARCH ON RARE DISEASES

1856. SHRI CONSTANDINE RAVINDRAN:

Will the Minister of **HEALTH AND FAMILY WELFARE** be pleased to state:

(a) whether Government has taken cognisance of recent advances in genetic research highlighting variations in clinical manifestations of rare diseases arising from mutations in the same gene;

(b) the measures introduced for strengthening genetic diagnosis, genomic testing, early identification and personalised management of rare diseases through specialised healthcare institutions;

(c) the assistance provided for genomic research and rare disease diagnostics;

(d) the collaboration with medical research institutions and biotechnology organisations for advancing precision medicine; and

(e) the future plans for improving access to affordable diagnosis and treatment for patients with rare diseases?

ANSWER
THE MINISTER OF STATE IN THE MINISTRY OF HEALTH AND FAMILY WELFARE
(SHRI PRATAPRAO JADHAV)

(a): Indian Council of Medical Research (ICMR) has informed that different variants within the same gene can produce a wide spectrum of clinical manifestations, as revealed by large-scale genomic sequencing. Further, different mutations within the same gene may result in distinct disease severity because they differentially affect protein function.

The Department of Biotechnology (DBT) has informed that it has taken cognisance of advances in genetics and genomics research through Mission Program on Pediatric Rare Genetic Disorders (PRaGeD), GenomeIndia Program and Unique Methods of Management of Inherited Disorders (UMMID) Program. The Biotechnology Research and Innovation Council-Centre for DNA Fingerprinting and Diagnostics (BRIC-CDFD), Hyderabad, has developed Indian Variant Database (INDVar), as a national repository of curated genomic and phenotypic information. The database

also enlists the variations in clinical manifestations arising from mutations in the same gene spread across the country, enabling clinicians and researchers to collaborate and improve diagnostic accuracy.

(b): Under the National Policy for Rare Diseases (NPRD), 2021, Government has designated 16 Centers of Excellence (CoEs), which are premier Government tertiary hospitals. As per the policy mandate, the CoEs are responsible for education & training at all levels, screening, diagnosis, prevention, treatment of rare diseases and research in the area of low-cost diagnostics & therapeutics. The COEs are given financial support up to ₹5 cr. for procurement of equipment as per individual center′s need for strengthening patient care services.

The DBT has implemented UMMID Program to strengthen genetic diagnosis and management. The program has established National Inherited Diseases Administration (NIDAN) Kendras for genetic diagnosis, prenatal testing, newborn screening and genetic counselling, along with Outreach and Training Centres. Nearly 2.75 lakh women and children have benefitted through screening and clinical genetic services.

Under Mission PRaGeD Program, DBT has focused to strengthen the diagnosis and management of rare genetic disorders in India. The program has established a nationwide network of clinicians for systematic workflow including patient recruitment, clinical phenotyping, genetic counselling, and genomic testing. Standard operating procedures for sample collection, sequencing, data analysis, and clinical reporting have been implemented to ensure uniformity and quality across the participating institutions. PRaGeD combines genomic testing with multidisciplinary clinical evaluation and functional characterisation of variants, particularly variants of uncertain significance (VUS), using advanced cellular and animal model systems. PRaGeD plays a key role in training the healthcare professionals for accurate genetic testing and diagnosis using workshops, lectures, webinars, conferences and other training modules across the country.

(c): DBT initiated the PRaGeD in project mode for 5 years duration in 2022 for Research & Development in areas of Human Genetics. PRaGeD aims to create awareness, achieve genetic diagnosis, discover & characterize new genes/mutations/variants, provide counselling, and to develop new therapies for pediatric rare genetic diseases of Indian population. In this project, 16 institutions are involved and an integrated workflow of comprehensive genomic sequencing with detailed phenotypic evaluation is achieved by the collaborative efforts of clinicians and research institutions across India. A total of 200 novel variants and 112 variants of uncertain significance (VUS) are identified. A web resource portal has been developed for the documentation of clinical phenotypes and identified genetic variants, leading to the establishment of mutation spectra of rare diseases in the country.

The GenomeIndia Program is a national scientific initiative to sequence genome of over 10,000 individuals representing India's diverse populations and build a local reference genome. The program has identified nearly 180 million genetic variants, including millions of rare and novel variants, providing an important resource for understanding genotype-phenotype relationships and rare genetic diseases in Indian populations.

ICMR also actively supports research initiatives and facilitates evidence generation in the field of rare diseases and other medical conditions. The medical research institutions and biotechnology organizations are funded for research in rare diseases aimed at understanding genotype–phenotype correlations, identification of novel disease-causing variants, identification of biomarkers, development of newer and cheaper diagnostics and development of therapies for rare diseases.

(d): Mission PRaGeD is a nationwide consortium of 16 institutions with expertise in clinical genetics, genomics, bioinformatics, molecular diagnostics, and functional biology, enabling multidisciplinary research. The collaborative network promotes translational research by connecting clinicians with basic scientists for functional validation of disease-associated variants.

The GenomeIndia Program is implemented by DBT through a consortium of leading academic and medical institutions across the country and has established national sequencing, biobanking and bioinformatics infrastructure for genomics research. The UMMID Program operates through Government medical institutions, NIDAN Kendras, Outreach and Training Centres to strengthen clinical genetics and genetic disease diagnosis.

(e): The Ministry of Finance provides exemption from Basic Customs Duty (BCD) and Integrated Goods and Services Tax (IGST) on the Drugs, Medicines and Food for Special Medical Purposes (FSMP) imported for persons suffering from rare disease for personal use or through CoEs till 31.03.2029. Further, NITI Aayog constituted a committee on "Drugs and Dosage Forms for Rare Diseases: Engagement with Manufacturers" to consider identification of a set of priority disorders / indications and their corresponding treatments that can be enabled for domestic manufacturing of orphan drugs. Indigenous development and marketing of drugs for select rare diseases has substantially lowered the costs to the extent that some drugs are available in the Indian market at 1/10th of the price of the imported Reference Listed Drug (RLD).

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