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

LOK SABHA
STARRED QUESTION NO. 111
TO BE ANSWERED ON THE 9TH FEBRUARY, 2018
GENETIC DISEASES

***111. SHRI SUNIL KUMAR SINGH:**
SHRI B. VINOD KUMAR:

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

(a) whether a large number of people in India are suffering from rare genetic diseases including Sickle Cell Anaemia and Thalassemia especially in tribal areas and if so, the details thereof along with the number of people suffering from rare genetic diseases in the country;

(b) whether the diagnosis rate of genetic diseases is extremely low, if so, the details thereof and the reasons therefor;

(c) whether the Government has identified the lacunae in public support system with regard to rare genetic disorders and if so, the details thereof;

(d) whether the Government proposes to bring out vaccine for any of such diseases, if so, the details including the estimated cost thereof along with the time by which it will be made available to the patients; and

(e) the necessary steps taken by the Government for early detection, treatment, promotion of research and creation of awareness about such diseases?

ANSWER
THE MINISTER OF HEALTH AND FAMILY WELFARE
(SHRI JAGAT PRAKASH NADDA)

(a) to (e) : A Statement is laid on the Table of the House.

STATEMENT REFERRED TO IN REPLY TO LOK SABHA STARRED QUESTION NO. 111* FOR 9TH FEBRUARY, 2018

(a) It is estimated that globally around 6000-8000 rare diseases exist. However, 80% of all rare disease patients are affected by approximately 350 rare diseases. So far about 450 rare diseases have been recorded in India from tertiary care hospitals. Lack of epidemiological data on incidence and prevalence of rare diseases impedes understanding of the extent of the burden of rare diseases in India. Recognizing the need to gather epidemiological data, Indian Council of Medical Research has recently launched 'Indian Rare Disease Registry' for certain rare diseases, including Thalassemia and Sickle Cell Anemia.

An estimated number of 1,20,000 to 1,50,000 patients are affected by Sickle Cell Disease in India with 20-35% carrier rate in tribals and other backward communities. The estimated number of beta Thalassemia cases is 1,00,000 to 1,20,000 with a carrier rate of 3-4% in overall population.

(b) Diagnosis facilities are available for screening, control and management of Hemoglobinopathies (Thalassemia, Sickle Cell disease) disorders. However, no study has been conducted on the diagnosis rate of other rare diseases in India. International data indicates that diagnosis of a rare disease may take upto several years. According to a recent report, it takes patients in the United States (USA) an average of 7.6 years and patients in United Kingdom (UK) an average of 5.6 years to receive an accurate diagnosis.

(c) Rare diseases poses a challenge to public health systems globally and more so in developing countries like India. Lack of epidemiological data, difficulty in conducting pan India epidemiological studies for each rare disease, challenges in diagnosis and management of rare diseases, challenges in research and development of drugs and diagnosis modalities, unavailability of treatment, the exorbitant cost of drugs for treating rare diseases etc. are a big challenge.

(d) Majority of rare diseases are inherited diseases and not treatable by vaccine. Also no vaccines are currently available for treatment of rare diseases. Some diseases like Sickle Cell anemia may be benefitted by pneumococcal vaccine in addition to the vaccines routinely recommended. These vaccines are provided free of cost by the States.

(e) Comprehensive Guidelines for prevention and management of Hemoglobinopathies have been developed and shared with states. ICMR, through

its flagship projects like Jai Vigyan has covered all aspects of overall diagnosis, training, awareness and management of Hemoglobinopathies in India and through the tribal health forum projects, the same has been extended to specific tribal groups in Maharashtra, Gujarat and Madhya Pradesh. ICMR and other Government funding agencies also promote active research in these areas through various intramural and extramural research programmes.

The Ministry of Health and Family Welfare has also formulated a National Policy for treatment of Rare Diseases in India to progressively build India's capacity to respond comprehensively to rare diseases. On the whole, the Policy seeks to strike a balance between the interest of patients of rare diseases and health system sustainability. ICMR has established 2 Centres of excellence of primary immune-deficiencies (PIDs) one at PGIMER, Chandigarh and one at ICMR-NIM, Mumbai for improving diagnostic facilities for PIDs and understanding their prevalence and pattern in India. Under NHM, States/UTs are also being supported for prevention and control and management of Thalassemia & sickle cell.

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