Rare Diseases – Challenges Associated with Rare Diseases

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Why in the News?

- Indian drug companies have started manufacturing medicines for at least four conditions, bringing down the cost by up to 100-fold.

About Rare Diseases

- A rare disease is a health condition of low prevalence that affects a small number of people compared with other prevalent diseases in the general population.
- There are 7,000-8,000 classified rare diseases, but less than 5% have therapies available to treat them.
- A condition is considered to be a rare disease if its prevalence is less than one case per 1,000 population.
- Around 6% to 8% of the population is estimated to have a rare disease, meaning 8.4 crore to 10 crore Indians are living with these conditions for which treatments either do not exist or therapies are extremely expensive.
- While a majority of rare diseases are believed to be genetic, many — such as some rare cancers and some autoimmune diseases — are not inherited.

Challenges Associated with Rare Diseases

- The field of rare diseases is complex and heterogeneous as prevention, treatment and management of these diseases has multiple challenges.
- Early diagnosis is a major challenge owing to a variety of factors that include lack of awareness among primary care physicians, lack of adequate screening and diagnostic facilities etc.
- Relatively little is known about the pathophysiology or the natural history of majority of rare diseases, particularly in the Indian context.
- Rare diseases are also difficult to research upon as the patients pool is very small and it often results in inadequate clinical experience.
- Despite progress in recent years, there is a need to augment effective and safe treatment for rare diseases.
- The cost of treatment of rare diseases is prohibitively expensive. For example, treatment for Spinal Muscular Atrophy (SMA) costs approximately Rs. 16 crore.
- Various High Courts and the Supreme Court have also expressed concern about lack of a national policy for rare diseases.

National Policy for Rare Diseases, 2021

- In 2021, the Union Ministry of Health and Family Welfare had approved the National Policy for Rare Diseases.
- Key Features of the Policy:
    - A provision for financial support up to Rs. 20 lakhs under the umbrella scheme of Rastriya Arogya Nidhi is provided for treatment, of those rare diseases that require a one-time treatment.
    - Objective: To lower the high cost of treatment for rare diseases with increased focus on indigenous research.
    - Eligibility: Beneficiaries for financial assistance are not be limited to below poverty line (BPL) families, but extended to about 40% of the population, who are eligible under Ayushman Bharat – Pradhan Mantri Jan Arogya Yojana (AB-PMJAY).
    - The policy has categorised rare diseases into three groups:
        - Diseases amenable to one-time curative treatment;
        - Diseases requiring long term or lifelong treatment;
        - Diseases for which definitive treatment is available but challenges are to make optimal patient selection for benefit.

- The policy envisages creation of a national hospital based registry of rare diseases.
- The policy focuses on early screening and prevention through primary and secondary health care infrastructure.
    - Screening is supported by Nidan Kendras set up by Department of Biotechnology.
- Policy also aims to strengthen tertiary health care facilities for prevention and treatment of rare diseases.
    - This is ensured by designating eight health facilities as Centre of Excellence (CoE).
    - These CoEs are provided with one-time financial support of up to Rs. 5 crores for upgradation of diagnostics facilities.

News Summary

- The Union government has announced a special initiative for manufacturing four types of homegrown ‘Made in India’ drugs for rare diseases for the first time.
- The government has prioritised 13 rare diseases and sickle cell disease.
- Currently, there are eight types of generic drugs and among them, four types of drugs are available in India and another four types of drugs will be available next year.
- The available generic drugs are for Tyrosinemia Type, Gaucher’s Disease, Wilson’s Disease and Dravet or Lennox Gastaut Syndrome- seizures.
    - Tyrosinemia Type:
        - Tyrosinemia type I is a rare autosomal recessive genetic metabolic disorder characterized by lack of the enzyme fumaryl acetoacetate hydrolase (FAH), which is needed for the final break down of the amino acid tyrosine.
        - The available priority drugs like Nitisinone (capsules) are used for the treatment of Tyrosinemia Type 1 and the cost difference of this drug is Rs. 2 crore per annum if purchased from other countries but an Indian company is manufacturing this in Rs. 2.5 lakh per annum.
    - Gaucher’s Disease:
        - Gaucher (go-SHAY) disease is the result of a buildup of certain fatty substances in certain organs, particularly your spleen and liver.
        - The drug Eliglustat recommended for the treatment of Gaucher’s Disease will cost India Rs. 3-6 lakhs per annum.
    - Wilson’s Disease:
        - Wilson disease (hepatolenticular degeneration) is a rare, autosomal recessive disorder caused by abnormal copper accumulation in the body particularly involving the brain, liver, and cornea.
    - Lennox Gastaut Syndrome:
        - Lennox-Gastaut syndrome (LGS) is a rare but severe form of childhood epilepsy characterized by a triad of multiple seizure types, characteristic EEG findings, and intellectual impairment.
- The government is working on a production-linked incentive scheme for these medicines.
- The medicines, at present, are available at the centres of excellence for rare diseases.
- In addition, the government is looking at whether making it available at Jan Aushadhi stores will be feasible.

Q1) What are Genetic Disorders?

Genetic disorders occur when a mutation affects your genes. Carrying the mutation doesn’t always mean you’ll end up with a disease.

Q2) What are Non Communicable Diseases?

The term NCDs refers to a group of conditions that are not mainly caused by an acute infection, result in long-term health consequences and often create a need for long-term treatment and care. These conditions include cancers, cardiovascular disease, diabetes and chronic lung illnesses.

Source: Major drop in cost of drugs for 4 rare diseases after Indian companies begin production | HT