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RARE DISEASE DAY

THIS ARTICLE COVERS 'DAILY CURRENT AFFAIRS' AND THE TOPIC DETAILS OF "RARE DISEASE DAY". THIS TOPIC IS RELEVANT IN THE "GOVERNANCE AND SOCIAL JUSTICE" SECTION OF THE UPSC CSE EXAM.

WHY IN THE NEWS?

Rare Disease Day was recently marked on February 28th. This international awareness day aims to raise awareness about rare diseases and their profound impact on patients and their families.

WHAT IS A RARE DISEASE DAY?

Rare Disease Day is an annual observance dedicated to raising awareness about rare diseases and their impact on the lives of individuals and their families. Celebrated on the last day of February each year, Rare Disease Day aims to highlight the challenges faced by those living with rare diseases and advocate for better support, research, and understanding of these often overlooked conditions.

THE OBJECTIVES OF RARE DISEASE DAY INCLUDE:

Educating and Uniting:

The day serves as a crucial platform to educate various stakeholders, including the public, healthcare professionals, policymakers, and researchers, about the existence, impact, and unique needs associated with rare diseases. This fosters a deeper understanding and fuels the creation of a more supportive environment.

Empowering Through Shared Experiences:

Rare Disease Day empowers individuals and families directly affected by these conditions. It provides them with a space to share their stories, experiences, and challenges, fostering a sense of community and solidarity among those facing similar journeys.

Fueling Research and Progress:

Through various campaigns and events, Rare Disease Day advocates for increased research funding and collaboration. This aims to accelerate advancements in understanding, treatment options, and potential cures for rare diseases. It's important to remember that while individual occurrences may be rare, the collective impact of these illnesses is significant, and research advancements can also benefit broader areas of medicine.

Improving Diagnosis and Care:

Efforts are continuously made to improve the diagnosis and treatment landscape for rare diseases. Raising awareness among healthcare professionals, advocating for better diagnostic tools, and ensuring timely access to specialised care are all crucial aspects of this ongoing process.

Building a Network of Support:

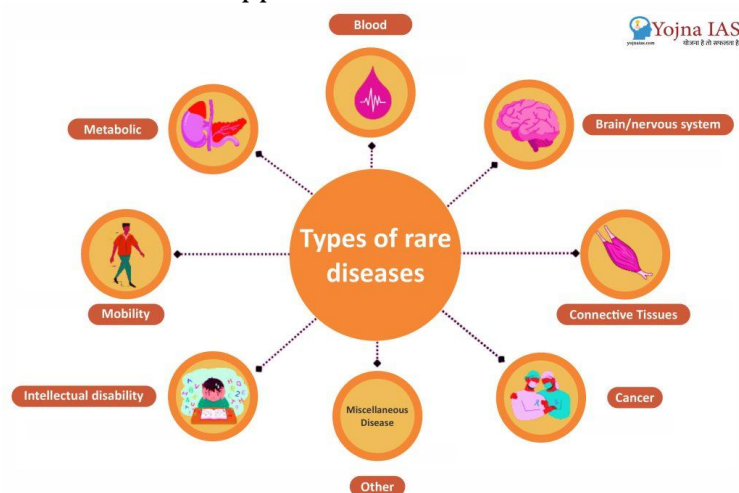
Rare Disease Day emphasises the importance of fostering connections and building support networks between individuals, families, and organisations working towards shared goals. These networks offer crucial emotional support, serve as valuable channels for information sharing, and provide a united voice in advocating for the needs of those affected by rare conditions.

Influencing Policy and Shaping the Future:

Advocates leverage Rare Disease Day to draw attention to the unique challenges faced by individuals with rare diseases. This ultimately aims to influence policy changes that improve their quality of life. These changes may involve addressing healthcare access, insurance coverage, and research funding allocations, paving the way for a more inclusive and supportive future.

ABOUT RARE DISEASES

- Rare diseases, alternatively termed orphan diseases, are medical disorders that impact a limited portion of the population. These diseases are characterised by their low prevalence, often impacting only a small number of individuals. While each rare disease may be uncommon on its own, collectively, it affects millions of people worldwide.
- The **World Health Organization (WHO)** defines rare diseases as medical conditions with a prevalence rate of **1 or fewer cases per 1,000 individuals**.
- **Rare diseases in India:** India accounts for **one-third of the worldwide instances of rare diseases, comprising more than 450 identified conditions**. Despite this substantial prevalence, rare diseases receive minimal attention in India, characterised by limited awareness, diagnosis, and drug development. An estimated 8 to 10 crore Indians are believed to be affected by rare diseases, with children constituting over 75% of this population. Unfortunately, less than 50% of the identified rare diseases in India have available treatments, and only around 20 diseases have approved treatments.



THE DAUNTING CLIMB: CHALLENGES IN FIGHTING RARE DISEASES

LIMITED AWARENESS AND UNDERSTANDING:

1. **Public knowledge:** The general public often lacks awareness about the vast spectrum of rare diseases, their symptoms, and the impact they have on individuals and families. This lack of understanding can lead to misdiagnosis, delayed intervention, and social stigma.
2. **Healthcare professionals:** Due to the rarity of these conditions, healthcare professionals may not have the necessary experience or expertise to accurately diagnose and manage them. This can lead to misdiagnosis, inappropriate treatment approaches, and frustration for patients seeking effective care.

DIFFICULTIES IN RESEARCH AND DEVELOPMENT:

1. **Small patient pool:** The limited number of patients with each rare disease makes it challenging to conduct clinical trials and gather sufficient data for research and development efforts. This can hinder the development of effective treatments and therapies.
2. **High costs:** Researching and developing treatments for rare diseases can be extremely expensive due to the smaller market size and the complex nature of these conditions. This can discourage pharmaceutical companies from investing in areas with limited financial return, leaving patients with fewer treatment options.

DIAGNOSTIC HURDLES:

1. **Non-specific symptoms:** Many rare diseases present with non-specific symptoms that overlap with more common conditions. This can make it difficult for healthcare professionals to pinpoint the underlying cause and lead to delayed diagnosis.
2. **Lack of readily available diagnostic tests:** Due to the rarity of the diseases, specific diagnostic tests may not be readily available or covered by insurance, further complicating the diagnosis process and leading to patient frustration.

ACCESS TO TREATMENT AND CARE:

1. **High cost of treatments:** Even when treatments are available, their high cost can create significant financial burdens for patients and their families. This can limit access to necessary therapies and impede their ability to manage their condition effectively.
2. **Limited availability of specialists:** Due to the rarity of these conditions, specialists with expertise in specific rare diseases may not be readily available in all regions, creating geographical barriers to accessing appropriate care.

GOVERNMENT INITIATIVES FOR RARE DISEASES IN INDIA

Recognising the challenges faced by individuals living with rare diseases, the Indian government has implemented several initiatives to improve their access to diagnosis, treatment, and support. Here's an overview of these key efforts:

- **National Policy for Rare Diseases 2021 (NPRD-2021):** This comprehensive policy serves as a roadmap for addressing various aspects of rare diseases in India. Some of its central features include:

1. **Categorisation of Rare Diseases:** The policy classifies rare diseases into three distinct groups based on treatment needs and associated costs:
 - **Group 1:** Diseases amenable to one-time curative treatment.
 - **Group 2:** Diseases requiring long-term or lifelong treatment with relatively lower costs.
 - **Group 3:** Diseases with available definitive treatment but with very high costs and lifelong therapy requirements.

- **Establishing Support Infrastructure:**
 1. **Centres of Excellence (CoEs):** Twelve CoEs have been established across India to provide specialised diagnosis, prevention, and treatment services for individuals with rare diseases.
 2. **Nidan Kendras:** These centres offer genetic testing and counselling services, playing a crucial role in the early detection and management of rare conditions.

- **Financial Support:** Under NPRD-2021, provisions are in place to offer financial assistance of up to Rs. 50 lakhs to patients suffering from any category of rare disease, facilitating their treatment at designated CoEs.

PRELIMS PRACTISE QUESTIONS

Q1. What financial assistance is offered to patients under NPRD-2021 for rare disease treatment?

- (a) Up to Rs. 10 lakhs
- (b) Up to Rs. 25 lakhs
- (c) Up to Rs. 50 lakhs
- (d) Up to Rs. 75 lakhs

Answer: C

Q2. According to NPRD-2021, how are rare diseases categorised based on treatment needs and costs?

- (a) Low-cost and high-cost categories
- (b) Curative, preventive, and lifelong treatment categories
- (c) Common, uncommon, and rare categories
- (d) Group 1, Group 2, and Group 3 categories

Answer: D

MAINS PRACTISE QUESTIONS

Q1. Examine the potential impact of artificial intelligence and machine learning on the diagnosis, treatment, and research landscape of rare diseases. Discuss both the potential opportunities and challenges associated with these advancements.

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