P.L.RAJ IAS & IPS ACADEMY
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1. UMMID Programme - Addressing Rare Genetic Disorders in India – Science & Technology

1. Why in News

1. Union Minister for Science and Technology dedicated UMMID (Unique Methods of Management of Inherited Disorders) Programme for Rare Genetic Disorders to nation
2. Initiative of Department of Biotechnology (DBT) improving diagnosis, management of rare genetic disorders in India
3. Seeks to make diagnosis, screening, counselling for inherited disorders affordable, accessible, widely available
4. Supports clinician training, institutional capacity-building in genetic healthcare
5. UMMID Dashboard launched, UMMID Compendium released strengthening nationwide monitoring, diagnostics, outreach services

2. About UMMID Programme

Objectives

1. Improving diagnosis and management of rare genetic disorders
2. Making diagnosis, screening, counselling affordable, accessible, widely available
3. Supporting clinician training (genetic counselors, medical geneticists)
4. Institutional capacity-building in genetic healthcare (laboratories, diagnostic centers)

Components

1. **UMMID Dashboard -** Digital platform for nationwide monitoring, tracking cases, diagnostics, outreach services; real-time data on rare disease patients
2. **UMMID Compendium -** Comprehensive resource documenting rare genetic disorders, diagnostic protocols, management guidelines for healthcare providers

Implementation

1. Department of Biotechnology (DBT) coordinating with medical institutions, research centers
2. Establishing diagnostic facilities, training programs
3. Strengthening referral networks connecting primary healthcare to specialized centers

3. What are Rare Genetic Disorders?

Definition

1. Diseases occurring due to abnormalities or mutations in genes or chromosomes
2. Generally inherited from parents
3. May affect physical, neurological, metabolic, developmental functions
4. Although each disorder affects small number, collectively impose major public health burden worldwide

Causes

1. **Inherited mutations in genes -** Major cause; passed from parents to children through DNA
2. **Chromosomal abnormalities -** Extra, missing, irregular chromosome portions leading to developmental, inherited disorders (Down syndrome, Turner syndrome)

3. **De novo mutations -** New mutations occurring spontaneously (not inherited)

Examples

1. **Sickle Cell Disease -** Hereditary blood disorder affecting red blood cell shape, functioning (crescent/sickle-shaped cells blocking blood flow, causing pain, organ damage)
2. **Thalassemia -** Inherited blood disorder reducing body's ability producing healthy hemoglobin (oxygen-carrying protein); causes anaemia, fatigue
3. **Haemophilia -** Genetic disorder impairing blood clotting process; prolonged bleeding even from minor injuries
4. **Duchenne Muscular Dystrophy (DMD) -** Genetic disease-causing progressive muscle degeneration, weakness; primarily affects boys
5. **Spinal Muscular Atrophy (SMA) -** Rare inherited disease damaging nerve cells controlling muscle movement; leads to muscle wasting, weakness

4. Challenges Associated with Rare Genetic Disorders

Delayed Diagnosis

1. Lack of awareness among healthcare providers, families
2. Inadequate genetic testing facilities (limited laboratories, equipment)
3. Many patients spend years consulting multiple doctors before accurate diagnosis
4. Misdiagnosis common due to symptom overlap with common diseases

High Cost of Treatment

1. Treatment, gene therapies expensive, unaffordable for most families (costs running into lakhs/crores)
2. Dependence on imported medicines further increasing healthcare expenditure
3. Long-term management requiring continuous medication, supportive care

Limited Healthcare Infrastructure

1. Limited number of specialized centers for genetic diagnosis, counselling (concentrated in metros - Delhi, Mumbai, Bengaluru, Chennai)
2. Advanced healthcare facilities for rare diseases mainly in urban areas
3. Rural, remote areas lacking access

Shortage of Skilled Professionals

1. Shortage of trained genetic counselors, clinicians, genomic researchers
2. Medical curriculum inadequately covering genetics, rare diseases
3. Brain drains - Trained professionals migrating abroad

Social Burden

1. Families affected face emotional stress (uncertainty, caregiving burden)
2. Social stigma (discrimination, isolation)
3. Financial hardship (job loss, catastrophic health expenditure)

Limited Research and Data

1. Lack of comprehensive data, disease registries affecting effective policymaking, research
2. Rare diseases receiving comparatively lower investment in scientific research, drug development (pharmaceutical companies focusing on common diseases with larger markets)
3. Missing epidemiological data on prevalence, distribution

5. Government Initiatives

National Policy for Rare Diseases (NPRD), 2021

1. Provides framework for prevention, management of rare diseases

2. Financial assistance for treatment of selected rare diseases requiring one-time therapies (up to ₹50 lakh under Umbrella Scheme Rashtriya Arogya Nidhi)
3. Three disease groups based on treatment availability, cost
4. Crowdfunding encouraged for expensive therapies

Rashtriya Bal Swasthya Karyakram (RBSK)

1. Provides early screening, intervention services for children with birth defects, developmental disorders
2. Screening at birth, 6 weeks, 6 months, 9 months, 2 years, 5 years
3. Covers 4 Ds - Defects at birth, Diseases, Deficiencies, Development delays

Sickle Cell Anaemia Elimination Mission

1. Government aims to eliminate sickle cell anemia as public health problem by 2047
2. Screening 7 crore people in 0-40 years age group in tribal areas (highest prevalence)
3. Awareness, counselling, treatment support

Other Initiatives

1. **Centres of Excellence -** Establishing specialized centers for rare disease diagnosis, treatment
2. **National Rare Disease Registry -** Creating database for epidemiological research, policy planning

6. Way Forward

Expand Screening Programmes

1. Expand new born, prenatal screening programmes across all states, districts
2. Universal new born screening for common genetic disorders (mandatory in all hospitals)
3. Carrier screening for high-prevalence disorders (thalassemia, sickle cell) in endemic areas

Establish Specialized Centers

1. Establish more specialized centers for genetic diagnosis, counselling (district-level genetic clinics)
2. Strengthen referral networks connecting primary healthcare to tertiary centers
3. Telemedicine for genetic counselling in remote areas

Invest in Research

1. Greater investment in biotechnology research, genome sequencing, indigenous therapies
2. Promote indigenous drug development reducing dependence on imports
3. Incentivize pharmaceutical companies for orphan drug development

Capacity Building

1. Train more genetic counselors, medical geneticists, laboratory technicians
2. Integrate genetics into medical, nursing curricula
3. Continuous medical education programs

Financial Support

1. Expand financial assistance coverage under NPRD
2. Include rare disease treatment under health insurance schemes (Ayushman Bharat)
3. Subsidize diagnostic tests, medicines

Awareness and Support

1. Public awareness campaigns reducing stigma
2. Patient support groups providing emotional, financial assistance
3. Advocate for rare disease patients' rights

7. Conclusion

UMMID Programme dedicated to nation improving diagnosis, management of rare genetic disorders through affordable, accessible screening-counselling, clinician training, capacity-building with

UMMID Dashboard-Compendium strengthening monitoring-diagnostics. Rare genetic disorders—caused by inherited mutations, chromosomal abnormalities (examples - Sickle Cell, Thalassemia, Haemophilia, DMD, SMA)—impose major public health burden. Challenges include delayed diagnosis due to limited testing, high treatment costs, limited infrastructure concentrated in urban areas, shortage of skilled professionals, social burden, limited research-data. Government initiatives—NPRD 2021 providing financial assistance, RBSK screening children, Sickle Cell Elimination Mission targeting 2047—complemented by UMMID. Way forward requires expanding new born-prenatal screening nationwide, establishing specialized centers, investing in biotechnology research-genome sequencing-indigenous therapies, capacity building, financial support expansion ensuring comprehensive, equitable genetic healthcare accessible to all Indians.

Source - **https -//newsonair.gov.in/union-minister-dr-jitendra-singh-dedicates-ummid-network-to-the-nation-at-a-function-in-new-delhi/**