NPRD, 2021

National Policy for Rare Diseases, 2021, on Prevention and Control of Rare Diseases

There are between 7000 – 8000 rare diseases reported globally. Despite technological advancements, less than **5% have therapies** to treat them. As said in NPRD, 2021, "the therapies are **exorbitantly costly and not universally available and accessible**."

NPRD 2021 acknowledges that the RGD research and public health policies in India are at a **nascent stage**. There is a need to undertake **systematic epidemiological studies** to ascertain the number of people suffering from rare diseases in India. Additionally, we know little about the **pathophysiology or natural history** of these diseases.

Any policy on rare diseases should consider the need for the utmost judicious utilization of limited resources for maximizing the overall health outcomes for RGD patients. The best strategy to reduce the burden of rare diseases is to **prevent their occurrence**. Prevention can be done at multiple levels as described in NPRD 2021 below:

PREVENTION OF RARE GENETIC DISEASES

PRIMARY PREVENTION

Avoiding occurrence of the disease

AVOID PREGNANCY IN ADVANCED AGE

AVOID MARRIAGE/PREGANCY WITH CARRIERS

- Especially when they are monogenic disorder carriers

IDENTIFY A COUPLE AT RISK

- Based on previous sib or family history of that disorder

SECONDARY PREVENTION

Avoiding the birth of affected children

PRENATAL SCREENING

- Biochemical screening and ultrasonography for chromosomal disorders
- Targetted screening for monogenic disorders using Next-Generation Sequencing

PRENATAL DIAGNOSIS USING INVASIVE TESTING

- Targetted diagnosis for chromosomal abnormality and single-gene/enzyme by Chorionic Villus sampling and amniocentesis

NEWBORN SCREENING & EARLY POSTNATAL DIAGNOSIS

- Newborn babies are screened before symptoms manifest for Lysosomal Storage Disorders (LSDs), and Severe Combined Immunodeficiency (SCID)

TERTIARY PREVENTION (REHABILITATION)

Providing better provisions to rare disease patients who present at an advanced stage

DEVELOPMENTAL ASSESSMENT

VISUAL & HEARING AIDS

EARLY STIMULATION & BEHAVIOURAL THERAPY

EMOTIONAL AND PSYCHOLOGICAL SUPPORT

PHYSICAL THERAPY

FEASIBILITY OF THESE STRATEGIES

NPRD 2021 points out that primary prevention strategies like avoiding pregnancy in advanced age are unrealistic. Therefore, secondary prevention is the best strategy. For secondary screening and diagnosis, NPRD recommends "**a screening and diagnostic strategy wherein those pregnant women in whom there is a history of a child born with a rare disease and that rare disease diagnosis has been confirmed, would be offered prenatal screening test(s) through amniocentesis and/ or chorionic villi sampling**".

Early diagnosis of rare diseases is a challenge since we lack awareness among primary care physicians and adequate screening and diagnostic facilities. Therefore, **11 Centres of Excellence** have been identified by NPRD and will be involved in **screening (antenatal, neonatal) and diagnostics**.