Abstract Title: Management of Rare diseases in India

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Abstract: India has witnessed a spectacular progression the diagnosis and management of rare diseases in the last decade. The declaration of rare disease policy by the Government of India in 2021 further strengthened the cause and opens doors and hope for thousands of Indian patients in availing treatment for rare diseases (RD) . There are various areas which need discussion in this context : Awareness, diagnosis, screening, prevention, treatment & research particularly in therapeutics.

The awareness has definitely increased in health care professionals , public and government due to availability of professional courses in medical education, CMEs ,outreach programs and designation of 12 Centres of Excellence for rare diseases under the RD policy .

The most commendable progress has happened in the area of diagnostics and almost all advanced genomic testing is available in our country both in public and private sector, more in the latter.

Screening and prevention which include preconception, prenatal counseling / screening and newborn screening ,though in place at some centres but not universal and need significant strengthening . The need for the hour is to have comprehensive care centres with clear management pathways.

Availability and accessibility to treatment has significantly improved with the provisions under RD policy but the main concern is for disorders which require one time/ life long treatments having exorbitant costs

Research has shown significant progress with the wholehearted support from various national and international funding agencies. The areas include, genomics, proteomics, metabolomics, gene discovery and therapeutics. Major efforts are required for developing therapeutics in the country to make the treatment affordable.

Recent developments in this area namely CAR-T therapy and "made in India drugs for four rare diseases " Gaucher’s disease, Tyrosinemia type 1, Wilson’s disease, Dravet Lennox Gastaut syndrome with the efforts of NITI Ayog strengthens the belief that the accessibility and affordability to treatment will improve further in times to come.

There is a need for all stakeholders to join hands in strengthening “ CARE for the RARE .

Area of expertise: Medical Genetics : Areas of interest - Intellectual disability Lysosomal storage disorders, Newborn screening, Rare diseases