Rare Diseases in India

Prelims - *Current events of National & International importance and General Science.*

Why in News?

Indian Organization for Rare Diseases (IORD) reported that out of 300 million people with rare diseases in the world, about 90 million are in India.

- **Definition –** According to WHO, Rare disease is a lifelong disease with a prevalence of 10 or fewer per 10,000 population.
- According to Organization of Rare Diseases India (ORDI) defined rare disease is defined as a disease that affects 1 in 5,000 people.
- **Rare disease in children -** 80% of these conditions are genetic in origin and predominantly affect children, about 30% of the children don't live to see their 5th birthday.
- Fewer than 5% of rare diseases have treatments approved by the U.S. Food and Drug Administration.
- More than 95% of these conditions don't have a therapy or it may be impossibly expensive.
- **Reason for rare disease in India -** Real number is likely to be higher since our social practices include endogamy, the practice of marrying within a community.
- Genetic condition in that community, endogamous marriages will tend to preserve that condition instead of letting it die out.
- **Consequences –** Expensive treatment, non-suitable insurance policies,
- Compensatory treatment by companies (not a feasible way of obtaining treatment), forced to get crowd funding.
- **Creating awareness - Premarital counselling -** Scientists and health advocates have collaborated with communities to discourage marriages between carriers of the same genetic mutation.
- This helps reduce the risk of passing on hereditary conditions to children.
- **Awareness among doctors** - Doctors may never have encountered them and are often unaware of these diseases.
- **Inform registry –** often medical problem is not reported, which would be essential to help draft suitable policies to support patients.
- Needed to help patients connect with each other, and so that industry identifies market opportunities to develop suitable therapies.
- **GenTICS -** Developed by Tata Institute for Genetics and Society, Bengaluru, a gene database on rare genetic disorders".
- This is a valuable resource, since a user can choose from a list of symptoms to predict

the possible rare disease.

- The patient's family can then take the information to their doctor and seek support from a patient group.
- **Molecular Diagnostics Counselling Care and Research Centre (MDCRC) in Coimbatore** – Working on Duchenne muscular dystrophy (DMD), a disorder that affects only male children, with females being the carriers.
- MDCRC has done large-scale genetic screening across several districts of Tamil Nadu intending to detect the relevant mutations early and eventually eradicating DMD from the State.
- Similar efforts are required for many disorders around the country.

Reference

The Hindu| Rare diseases have a lot to gain from greater awareness