Championing for the Rare

IDEAS
INSTITUTE FOR DEMOCRACY AND ECONOMIC AFFAIRS

WORK IN PROGRESS WHITE PAPER:
Rare Diseases in Malaysia

Professor Dr Thong Meow Keong
Affiliation: Genetic Medicine Unit, Department of Paediatrics, Faculty of Medicine, University of Malaya

Dr Azlina Ahmad-Annuar
Affiliation: Department of Biomedical Science, Faculty of Medicine, University of Malaya

1. Definition and accurate data relating to rare diseases in Malaysia is needed:

    a. The lack of a national definition and accurate data on rare diseases lead to challenges in formulating a national health policy on the needs of individuals with rare diseases

    b. Agree to select an accurate and representative definition for rare disease in Malaysia and to enable comparative studies using an international standard terminology.

    c. Create and maintain an accurate and up-to-date national registry on rare diseases, including genetic and congenital conditions.

    d. Existing databases in Ministry of Health hospitals and university hospitals – issues of access to data, patient consent, long term sustainability of a registry, sharing of information and confidentiality issue, legal issue (PDPA Act)

2. Awareness and educational programs on rare diseases for patients, health professionals and general public:

    1) Commit to training more health professionals and genetic counsellors to help families with rare diseases and to avoid delay in diagnosis.

    b. Increase awareness amongst key stakeholders in Malaysia on rare diseases such as policymakers, academics, media organizations and non-governmental bodies.

    c. Networking, coalition building and media education to raise the profile and sensitization of the general public to the issue of rare diseases.

    d. Greater focus on the topic of rare diseases in medical training and curricula

    e. Effectiveness of current dissemination and awareness programs

3. Current governmental strategies and practice towards rare diseases

    a. Need for collaboration and involvement of all relevant agencies and ministries
        1. Lack of RD policy and national coordinating agency for RD
        2. Overlapping jurisdictions on RD amongst different Ministries
        3. Improve inter-ministry collaboration in treatment of rare disease.

    b. Review of current capabilities in government in terms of equipment and expertise
        1. Track record and experience of managing RDs in terms of programmes and their implementation

Review of funding structures

    a. Government: Adequate funding and resources must be allocated to provide access to diagnostic tests, treatment and rehabilitation to all patients with rare diseases and their families.

    b. Support from pharmaceutical companies for treatment at present

    c. Engaging the insurance industry to remove discrimination against patients with inherited rare diseases.

    d. Health economic models for RDs (refer to A/P Asrul IDEAS Policy) Current funding structure for drugs and therapies

5. Funding and expertise for treatment and rehabilitation services

    a. Increase the budget for investigations, treatment and management of rare diseases in all types of healthcare facilities and university hospitals to achieve equity of care and services regardless where the patient is domiciled.

    b. Increase funding for rehabilitation services, including equipment and more qualified therapists.

    c. Increase accessibility for disabled persons in general, which would also incorporate those patients with rare diseases.

    d. Provision of respite care for parents and caregivers of patients with rare diseases that caused disabilities.

    e. Improve palliative care services for end-of-life situations in patients with rare diseases.

6. Legislation is needed to protect the rights of individuals with rare diseases

    a. Issues about equity and equitable access to treatment; the ethical aspects and protection of rights of children and patients with RDs.

    b. Rare Diseases and Orphan Drug Act: Legislation is urgently needed to protect the rights and lives of babies, children and persons with rare diseases. This is in keeping with international standards and practices and will help develop specific criteria for evaluating new orphan drug listing in the Ministry of Health Formulary.

    c. Increase job opportunities and educational training and support for disabled persons with rare diseases.

7. Pre-pregnancy and Pregnancy Care

    a. Food fortification with folic acid to prevent neural tube defects.

    b. Educational programs for all women of childbearing age and healthcare providers regarding avoidance of teratogenic drugs and obesity.

    c. Educational programmes for sonographers, obstetricians, nurses and midwives to detect signs of possible rare disease during prenatal check-ups and upon birth.

    d. Educational programmes for nurses and midwives on how to provide immediate care for babies born with rare diseases, for example those with epidermolysis bullosa, osteogenesis imperfecta (brittle bone disease) and spinal muscular atrophy.

8. Newborn screening program for diseases that cause severe disabilities and death:

    a. Expand current newborn screening to provide prompt diagnosis and treatment to avoid death and/or disabilities.
        I. Newborn screening for critical congenital heart diseases,
        II. Inborn errors of metabolism
        III. Hearing loss
        IV. Rare primary immunodeficiencies,
        V. Neurological conditions such as spinal muscular atrophy and others.

    b. Access and mandatory screening programs nation-wide
    c. Availability of confirmation tests and early treatment

9. Clinical expertise and laboratory facilities to diagnose serious genetic diseases:

    a. Capacity-building in critical areas of expertise for rare diseases such as genomic scientists, clinical geneticists, genetic counsellors, genetic nurses and technologists and primary care providers.

    b. Create government posts for new areas of employment e.g. genetic counsellors and genomic scientists as well as data managers.

    c. Increased diagnostic laboratories for genetic and genomic diseases in various parts of the country.

10. Research and development in rare diseases as one of the priority areas

    a. More research funding is needed for rare diseases that may not be 'commercially' beneficial. Many new innovative pharmaceuticals and approaches to therapy are a result of new insights from research into rare diseases.

    b. Explore the possibilities for joint venture with pharma and biotechnology companies to produce biopharmaceuticals, biosimilar products, biosensors and medical devices locally, thus generating new industries in the field of rare diseases.