Executive Summary

The Second Malaysia Conference on Rare Disorders (the Conference) convened on the 25th and 26th of October 2013 at the Majestic Hotel, Kuala Lumpur with the theme "Developing Strategies for a National Rare Disease Plan". The Conference achieved two notable firsts. The Conference was a first-time coalition effort by three patient support groups, namely, the Malaysian Rare Disorders Society, the Malaysia Lysosomal Diseases Association and the Malaysia Metabolic Society. Secondly, it was the first time that participants representing important stakeholders gathered together to discuss the development of such a plan. The Conference received strong support and cooperation from the Ministry of Health, Malaysia.

The challenges faced in Malaysia in regard to rare diseases are:

- late treatment as a result of late diagnosis or misdiagnosis
- inaccessibility of genetic services
- high drug costs, limited funding for treatment, testing and research in rare diseases
- inadequate and unequal access to services between urban and rural hospitals and between government hospitals and university hospitals
- lack of support services for rare disease patients such as rehabilitation services, welfare and employment opportunities
- lack of rare disease experts and trained support staff
- lack of local data on rare disease.

Hence the development of strategies for a national rare disease plan that can provide a holistic approach to providing care for patients with rare disease.

Four forums gave participants invaluable insight into the rare disease situation, both at international and local levels, right down to the management of rare disease patients, based on the following sub-themes:

- To develop strategies for a national rare disease plan
- To coordinate national activity on rare diseases
- To provide multidisciplinary clinical care and systematic approach for management of patients with rare diseases
- To ensure effective on-time treatment.

Papers by local and international speakers as well as presentations by patient support groups served as the reference for two roundtable discussions on the sub-themes.

KEY OUTCOMES

- A national rare disease plan is required. A plan will provide a framework covering clinical, public health and disability services that addresses prevention, timely diagnosis, early

intervention, adequate treatment and rehabilitation of patients with rare diseases. Towards this end:

- Patients with rare diseases must be officially recognised by the government and accorded priority status by the Ministry of Health.
- It is necessary to adopt a standard definition of rare disease for Malaysia to guide policy development, academia, research, disease education, diagnosis and screening, treatment and financial support.
- There must be a dedicated budget and funding for rare diseases in terms of treatment, training, facilities and research.
- A national patient registry will facilitate research and planning.
- A generic model of care or clinical pathway is necessary for good management of rare diseases which should include prenatal diagnosis and newborn screening for high risk groups, and effective on-time treatment.
- Solutions should be found to address the unequal access to care due to the gap in service delivery between urban and rural hospitals, and between government hospitals and university hospitals.

- A national alliance of rare disease organisations or groups in Malaysia should be established to speak with one voice and by its establishment, create a stronger voice to advocate for patients with rare diseases and their families. The national alliance will raise awareness of the impact of rare diseases, bring up issues affecting patients with rare diseases and their families and support the government in formulating rare disease-friendly policies.

- In the area of capacity building, rare disease studies should be included in the curriculum of medical and allied healthcare students at undergraduate and postgraduate level. In addition, the position of genetic counsellor should be created in the government service where there is currently none. The genetic counsellor has an important role to play as part of the genetic support team.

- Recognition and affirmation that rare disease patients have rights under the Universal Declaration of Human Rights, the Convention on the Rights of the Child (which Malaysia ratified in 1995) and under the Persons with Disabilities Act 2008, specifically the right to treatment. Further, individuals with rare diseases should be recognised as persons with disabilities by the relevant ministries.

- Matters raised during the Conference should be followed up by the establishment of a multi-stakeholder committee that brings together all stakeholders for engagement and consultation. It is suggested that the Ministry of Health is the most suitable party to take the lead.

In conclusion, the Second Malaysia Conference on Rare Disorders 2013 generated a consensus by the participants that a national rare disease plan is important and action should be taken to develop and implement it so that patients with rare diseases can receive the same opportunity to life and health, like any other individual.