PLENARY 2

NEW GENETICS AND ITS IMPLICATIONS FOR PAEDIATRIC PRACTICE

Thong Meow Keong

Genetics & Metabolism Unit, Department of Paediatrics, Faculty of Medicine, University of Malaya, Kuala Lumpur

Genetic disorders and birth defects collectively form an important component of the health burden and a main cause of chronic illnesses in childhood. Three percent of newborns have major birth defects and represents a major contributor to under-five mortality in Malaysia. While single gene disorders are important from the family’s perspective, other multifactorial conditions such as neural tube defects and cleft lip and palate involving the interplay between genetic and environmental factors are important from the public health’s perspective. In addition, conditions with strong genetic susceptibility such as cancers, diabetes and asthma have become increasingly prominent and many families are turning to their paediatricians advice and information.

Prevention of genetic diseases may be achieved through empowering individuals with information about their genetic risks and enabling them to make informed choices about their reproductive options. Reducing genetic morbidity and mortality would be a secondary goal of prevention. The paediatrician can assist the clinical geneticist in playing an active role in this respect. This include public education regarding genetic diseases, birth defects and strategies to avoid teratogens, providing basic genetic counselling for at-risk families, promotion of periconceptional folic acid supplementation and advising pre-pregnancy management of maternal illnesses and early antenatal visits once pregnancy is confirmed. These strategies require an understanding of the natural history of the disorders, genetic testing techniques and their limitations, and the basic knowledge of genetic counselling.

A good family history will help in identifying family members at risk for genetic diseases. Genetic counselling is defined as a communication process that deals with the human problems associated with the occurrence or risk of occurrence of a genetic disorder in a family. Genetic counselling helps the individual or family to understand the diagnosis, burden of disease and the available management, the risk of recurrence in relatives and makes the best possible adjustment to the disorder in an affected family. The paediatrician must be able to differentiate between merely giving genetic information and performing genetic counselling.

Providing timely information on the avoidance of teratogens and encouraging all women of child bearing age to consume periconceptional folic acid supplementation also reduce birth defects such as neural tube defects. Population screening and counselling for genetic diseases may empower carriers to make appropriate decision regarding the choice of life partners and prenatal diagnosis. Technology has also changed the roles of the paediatricians, for example accessibility to internet information and the introduction of newer techniques leading to over-medicalisation and raising parental anxiety are some of the implications in paediatric practice today. The paediatrician must be aware of the ethical, legal, social and religious implications of this ‘new genetics’.

The paediatrician’s evolving roles of this ‘new genetics’ will become increasingly crucial for the health of children in the decade to come.