ICASH-KEYNOTE

BRINGING MEDICAL GENETICS TO THE PUBLIC: THE CHALLENGES FOR INDONESIAN MEDICAL FACULTIES

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EXTENDED ABSTRACT

Medical genetics is one of the most rapidly advancing fields of medicine, and molecular genetics is now integral to all aspects of biomedical science. Every physician who practices in the 21st century must have an in-depth knowledge of the principles of human genetics and their application to a wide variety of clinical problems.

Millions are currently affected by hereditary diseases around the world. Statistical analysis showed about 5% of all pregnancies carries a child having abnormalities including genetic disorders, congenital malformation or significant disability. It is estimated that 43% of patients with mental retardation are caused by monogenic disorders or chromosomal anomalies. In developed countries, fully or partially inherited diseases account for about 45% of admission in paediatrics in hospitals. In developing countries, around 25% of prenatal and infant mortality are caused by hereditary diseases and most death-causing non-infectious disease probably have a genetic component.

Medical genetics is targeted to provide service to people with genetic or inherited conditions and their families with the final goal of helping individuals and their families to cope with their conditions and get the necessary support to live a life as normally possible. Currently, there are no medical school in Indonesia that specializes in genetics in the undergraduate level and with the population that ranked 4th worldwide, the amount of individuals affected by genetic and inherited conditions in Indonesia is very large and most of them are not identified due to underdiagnoses and lack-of-knowledge by the primary health care provider.

In the United States, medical genetics has been incorporated into medical school curriculum for a number of years. In 2001, the American Society of Human Genetics (ASHG) and the Association for Professors in Human and Medical Genetics (APHMG) issued the “Medical School Core Curriculum in Genetics”. In 2009, the American Association of Medical Colleges (AAMC) and the Howard Hughes Medical Institute (HHMI) formulated competency-based curriculum that were required for future physicians. Accreditation Council for Graduate Medical Education (ACGME) and American Board of Medical Specialists (ABMS) then emphasizes medical graduates on competencies such as, medical knowledge, patient care, interpersonal and communication skills, practice-based learning and improvement, professionalism, and

system-based practice. In 2013, the ACGME/ABMS broad competencies were used by the Association of Professors of Human and Medical Genetics (APHMG) to specifically apply the AAMC-HHMI standards to medical genetics education as well as to update their recommended genetics and genomics competencies. The resulting APHMG core-curriculum focuses on developing basic and applied empirical skills in genetics for medical students and emphasizes on making appropriate decisions on ordering particular molecular tests for patients, understanding how to use bioinformatics tools as part of diagnosis and treatment decisions, managing patient symptoms, and making appropriate referrals to specialists- ethically and professionally. The APHMG curriculum competencies includes, but not limited to: 1. Knowledge in genome organization/gene regulation, 2. Genetic variation, 3. Population genetics, 4. Principles of inheritance, 5. Cytogenetics and molecular genetics, 6. Biomedical genetics, and 7. Cancer genetics.

University of Rochester School of Medicine and Dentistry in the United Kingdom has genetics in medical school curriculum based on the 2001 ASHG/APHMG-recommended "Medical School Core Curriculum in Genetics" and believed that medical genetics must be explicitly included in the curriculum. Following their medical school curriculum reform in 1999, URSMD formulated the Double Helix Curriculum (DHC) which emphasizes on the "intertwining strands" of basic and clinical science education throughout their four-year curriculum. The medical genetics curriculum at URSMD was constructed to adhere to the ASHG/APHMG guidelines. The first year medical students were exposed to the basics of genetics such as taking family history, molecular genetics, the central dogma, population genetics and genetic anthropology, basic clinical genetics, and "omics". The integration continues to the second year with topics in neurogenetic disorders, pathogenesis of various genetic disorders, prenatal diagnosis and artificial reproductive technology, as well as the opportunity to experience practical clinical cytogenetics and practical molecular diagnostics. The third year of the curriculum includes integrative clinical genetics with topics in infertility, epigenetics, teratology, genetics of developmental pathways, and hands-on syndrome diagnosis exercise. The final year of the curriculum emphasizes on clinical genetics and writing a grant proposal for research involving genetic approaches.

At Faculty of Medicine Universitas Swadaya Gunung Jati (FMUGJ), integration of genetics into the 3.5-year pre-clinical and 2-year clinical medical curriculum will start shortly. The curriculum will include teaching of basic genetics in the first year. The second year follows up with genetic disease caused by mutations and chromosomal abnormalities. The third year will include dysmorphology, physical examination for genetic and inherited conditions, and updates on the latest cytogenetic and molecular techniques. The material will be delivered through a mixture of lectures, problem-based learning and practical skills lab.

In conclusion, medical genetics services must be established at all levels of health care and spear-headed by primary-care physicians. The challenge is to formulate a medical genetics curriculum that give genetics knowledge to future medical students to be functionally applied in a local and national settings. Therefore, it is very important for medical schools to integrate their curriculum with genetics in order to follow with the rapidly-expanding advancing field and answering the ever-growing challenge of diagnosis and providing care to those affected by genetic and inherited conditions.