

## Genetic counseling in Indonesia as a mandatory service

Lantip Rujito\*<sup>1</sup>

<sup>1</sup>Department of Molecular Biology, Faculty of Medicine, Universitas Jenderal Soedirman, Central Java

### GUEST EDITORIAL

#### ARTICLE INFO

\*Corresponding author:  
l.rujito@unsoed.ac.id

DOI : 10.20885/JKKI.Vol9.Iss1.art1

Copyright ©2018 Authors.  
This is an open access article distributed under the terms of the Creative Commons Attribution-NonCommercial 4.0 International Licence (<http://creativecommons.org/licenses/by-nc/4.0/>).

Genetic counselling is a comprehensive process undertaken by a genetic counsellor to a patient and or patient's family to a disease that is thought to be related to a particular genetic condition. This involves various parties such as molecular geneticist, paediatrician, neurologist, surgeon, or internist, by the facts and symptoms of any preventive, diagnostic, management, and prognostic approach.<sup>1</sup> The layman mentions that genetic counsellors are associated with rare diseases.

The global gene organisation stated that in general, rare diseases were not a priority in many national health programs in developing countries. Three hundred million people reflecting about 6-8% of the total world population is estimated to suffer from this kind of rare diseases. Currently, 7000 to 10000 different cases are documented around the world. It is counted that 30 million people in the United States are living with rare diseases. This equates to 1 in 10 Americans or 10% of the U.S. population. Similar to the United States, Europe has approximately 30 million people living with rare diseases.<sup>2</sup>

Indonesia itself is a country with complex genetic diversity. These facts are reflected in the ethnicity, linguistic, and cultural diversity which stretches from island to island. The process of migration and mixed marriage are two of the causes of diversity. What is about the epidemiology of rare disease in Indonesia? The exact number is hard to reveal since there is no official data from the government. However, from the research literature, we can estimate that Indonesia is potential to have a high rate of rare diseases. Carrier of thalassemia, one of the recessive disorders reaches 10% of the population.<sup>3</sup> Using the law of Hardy-Weinberg, it can be estimated that the number of thalassemia patients in Indonesia currently is 2 million patients, registered or not registered. With the birth rate of 4 million babies per year, affected babies born per year is approximately 40,000. Other diseases such as down syndrome also mentioned there are one among 700 people.<sup>4</sup> In Indonesia, extrapolating data found about 300,000 people is living with Down syndrome. Haemophilia, a disturbance of blood clotting has a ratio of 1 in 10,000 in developing countries.<sup>5</sup> The case of Fragile X Syndrome with mental retardation as a hallmark occurs in hereditary in the Semin Gunung Kidul area.<sup>6</sup> At first glance, it appears that the prevalence of the rare disease is very small if it is seen case by case. However, if this prevalence is combined, it shows an unusually large number. If all of the people with rare diseases live in one country, it would be the world's 3rd most populous country.<sup>2</sup> If converted to the population of Indonesia, 12.5 million people are living with a variety of rare diseases.

The problem of rare disease is very complicated. With the base of genetic mutations reaching 80%, the diseases are often carried from birth to the end of life. Rare diseases also account for 35% of deaths at an early age, and not to mention the enormous cost of therapy financing. Thalassemia studies in

Indonesia reported that both direct cost and indirect cost required approximately 10 million Rupiahs per month per patient, and this goes from birth to the death.<sup>3</sup> An ambiguous case of genitalia requires lifelong hormone treatment that is not supported by insurance and must be imported from another country. Other obstacles are cases of death such as cystic fibrosis in children, or early paralysis due to neurological disorders in cases of Duchene Syndrome. This kind of problem leads to super-expensive management, life-long treatment, and often poor prognostics result.

In some diseases, prevention is the right step to stop the increase of a rare disease. Recessive diseases, such as thalassemia which burdens super-expensive treatments can be prevented theoretically with super-easy prevention programs; avoiding unification of recessive genes, with a series of screening programs, premarital counselling, prenatal diagnosis and mass education for the community.<sup>7</sup> Providing knowledge and extensive education supported by carrier examination is believed to bring the benefit of the country in reducing the future of new cases. Early identification and registration of mutant gene carrier suspects can be applied to ensure comprehensive treatment, bringing targeted therapy precisely, and better future planning for patient. This task can be handled by a genetic counselor who has received a series of training and teaching about the genetic aspects of various diseases. Indonesia with a population of almost 250 million requires the presence of genetic counsellors as well as systemic referral patterns in the management of rare disease.<sup>8</sup> Regulation of genetic counsellor education, counsellor registration system, and spreading of genetic counsellors are the most important issues that must be embedded in the national health system. Without the attention and the first step to thinking about these rare diseases, the state is neglecting to pay attention to this communities.

## REFERENCES

1. Mester J, Schreiber A, Moran R. Genetic counselors: Your partners in clinical practice. *Cleveland Clinic Journal of Medicine*. 2012;79(8):560–8.
2. Global Genes. RARE diseases: Facts and statistics [Internet]. 2017. Available from: <https://globalgenes.org/rare-diseases-facts-statistics/>
3. Kementerian Kesehatan Republik Indonesia. Pencegahan thalassemia (Hasil kajian Health Technology Assesment tahun 2009). Jakarta; 2010.
4. Parker SE, Mai CT, Canfield MA, Rickard R, Wang Y, Meyer RE, et al. Updated national birth prevalence estimates for selected birth defects in the United States, 2004–2006. *Birth Defects Research Part A: Clinical and Molecular Teratology*. 2010;88(12):1008–16.
5. Alabek M, Mohan R, Raia M. Genetic counselling for hemophilia. 2015.
6. Faradz S, Buckley M, Lam-Po-Tang, Leigh D, Holden J. Molecular screening for fragile X syndrome among Indonesian children with developmental disability. *American Journal of Medical Genetics*. 1999;83(4):350–1.
7. Cao A, Kan YW. The prevention of thalassemia. *Cold Spring Harbor Perspectives in Medicine*. 2013;3(2):a011775.
8. Rujito L, Ghozali PA. Initiating development of genetic counseling services in the health services unit: An initial assessment. *Journal of the Indonesian Medical Association*. 2011;60(2):426–30.