TRANSLATIONAL MEDICINE IN INDONESIA

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Indonesia
Genomic research

- May improving:
  - The quality of diagnoses,
  - Identifying predisposition factors,
  - Detecting etiology,
  - Developing drugs adapted to genetic information etc.
The Indonesian genomic research progress can be observed through:

✓ Publications
✓ Funding of research
✓ Scientific property such as: dissertation, thessis
✓ Lectures.
Situation

- Infectious and degenerative diseases still dominate and use the largest resources and costs.
- Lack of data about prevalence of genetic diseases.
- Surveillance for genetic diseases has not been done systematically.
• Limited facilities are considered as the main issue in accelerating these aims.
• The high cost of genetic studies has also become a problem not only for patients but for researcher.
• Not many researchers are interested in this field because Genetic diseases were considered as an incurable.
Most of patient’s medical expenses now covered by government support with BPJS (government insurance)

• Genetic testing still expensive and there is a big number of patients

⇒ it is difficult for the government to cover all of costs.

⇒ Then financing priorities are for the main tool of diagnosis and treatment
Thalassemia in Indonesia

• is the major genetic problem in Indonesia.
• 2017: more than 8000 pts and >50% in Java island
• Its has specific characteristic due to the wide variability of the mutations.
• All diagnosis and treatment expenses cover by Government insurance
• Genetic testing not performed routinely, most only for research purposes or special cases.
Prevalence of carrier in Indonesia:

• The diversity of the genetic background can be seen in the carrier frequency of:
  - b-thalassemia (5–10%)
  - Hb E (1–33%)
  - a-thalassemia (6–16%)

➔ from the various ethnic population.

• This variation resulted to unequal anticipated carrier testing and prenatal testing workload. (Wahidiyat et al. 2006).
Genetic Laboratory

- Eijkman Institute and CEBIOR UNDIP are two of the leading research centers in Indonesia which also developed genetic laboratory.
- Now some universities also have genetic laboratory such as UNPAD Bandung, UGM Jogjakarta etc.
- Eijkman Institute initiated mainly for thalassemia became expanded for other diseases such as Spinal Muscular Atrophy (SMA), Duchenne Muscular Dystrophy (DMD), achondroplasia, Down Syndrome, Klinefelter syndrome, Turner syndrome, Disorder of Sexual Development (DSD) cluster, Prader-Willi syndrome and/or Angelman syndrome.
- Premarital and preconception counseling
- Prenatal diagnosis using amniocytes sample.
- DNA forensic, Disaster Victim Identification (DVI), and perpetrator identification
CEBIOR (Center for Biomedical Research) at faculty of Medicine, Diponegoro University, is also one of the leading centers providing genetic laboratory since 1999.

Genetic testing is mainly for cytogenetic and some of common genetic diseases caused by certain gene mutations.

Other genetic testing are being done such gene mutation for Fragile X syndrome, SRY and AZF gene microdeletion for infertility and genital defect cases etc.
Thalassemia Genomic studies in Indonesia

• Research in genetics and genomic medicine has been done sporadically in several research centers in Indonesia.
• The role of co-inherited of α-thalassemia and xmn^{γγ} polymorphism as clinical modifying factor in β-thalassemia with ivs1nt5 homozygote mutation (Lelani Reniarti, 2011)

• Influence of polymorphism gen hamp-p -582 a> g and hf h63d to iron status career talasemia Beta properties. (Nyoman suci 2012)

• Interaction of Hb adana (HBA2: c.179G>A) with deletional and nondeletional α(+)-thalassemia mutations: diverse hematological and clinical features. (Nainggolan IM 2013)
• A 65 bp deletion in band 3 gene of beta-thalassemia patients in Indonesia. (Dewajanthi 2014)

• Severe α-thalassemia intermedia due to a compound heterozygosity for the highly unstable Hb Adana (HBA2: c.179G>A) and a novel codon 24 (HBA2: c.75T>A) mutation. (Megawati 2015)

• Molecular Scanning of β-Thalassemia in the Southern Region of Central Java, Indonesia; a Step Towards a Local Prevention Program. (Rujito 2015)
• Hydrops fetalis associated with homozygosity for Hb Adana [alpha59(E8)Gly-->Asp (alpha2)]. (Nainggolan IM 2016)

• Modifying effect of XmnI, BCL11A, and HBS1L-MYB on clinical appearances: A study on β-thalassemia and hemoglobin E/β-thalassemia patients in Indonesia. (Rujito, 2016)
• Natural resistance-associated macrophage protein 1 gene polymorphisms in thalassemia patients with tuberculosis infection (Mohammad Ghozali et all 2016)

• Optimization of Pyrosequencing Method to Detect IVS1-NT5 β-Globin Gene Mutation in β-Thalassemia (Maskoen, 2017)

• Mutation Spectrum of β-Globin Gene in Thalassemia patients at Hasan Sadikin Hospital (Maskoen, 2017)
• Several collaborations with countries abroad have been done and should continue.
• CEBIOR Has MOU and collaboration with:
  - VUMC Amsterdam NL
  - Radboud Univ Nijmegen NL
  - ERASMUS univ Rotterdam NL
  - KK hospital singapore
  - Poitiers University
  - Mind Institute UC Davis California
Conclusion

• There is still a gap between genomic research and practices
• Genetic testing not cover by insurance then not routinely used for one of diagnosis tool.
• The universities together with government have responsibility to enhance the number of research and improving disease services
• Indonesia still has to accelerate this effort to be able to catch up this goal.
Thank YOU