CHAPTER VI
CONCLUSION AND FUTURE DIRECTIONS

VI.1 Conclusion

1. There is no SCN1A mutations in Indonesian mental retardation with epilepsy population.
   There are SNPs and UV found in SCN1A screening.

2. There is no ARX mutations in Indonesian mental retardation with epilepsy population.

3. There is no STXBP1 mutations in Indonesian mental retardation with epilepsy population.
   There are UVs found in STXBP1 screening.

4. There is no LGII mutations in Indonesian mental retardation with epilepsy population.

   The absence of genetic abnormalities in the coding region of the genes that was screened indicates that genes tested in this study in this population have very low frequency. Nevertheless, new unclassified variants in the patients list were found. Results also strengthen the importance of genetic counselling especially parental testing before conclude any mutation finding on sequencing method.

   Thus, present results suggest that genomic variations in screened genes are not related to susceptibility to MR with epilepsy, at least in the Indonesian population. Presented data and the findings of previous studies suggest that genetic factors other than mutation of SCN1A, ARX, STXBP1, and LGII genes are involved in the etiology of MR with epilepsy. Further studies of larger and different populations are needed.

VI.2 Future directions
From the patient’s side, more detailed clinical examination with brain scanning and EEG to determine specific type of seizures were needed, in order to get better differential diagnosis for each patient.

From the molecular side, other candidate genes to be sequenced can be chosen. Also, do the SNP array for all of the patients, to exclude the structural aberration. In the future, exome or whole genome sequencing may find the underlying genetic defect for each patient.

 Genetic counselling aspect is also very important. This research proved that parents testing has diagnostic strength, and may lead to the right conclusion. By doing more advance counselling techniques and giving continuous information may help all parties involved got the best result among all.

This research project raises the level of awareness about epilepsy mental retardation cases, so that researchers and clinicians can finally conclude the very fundamental molecular and clinical genetic base for those patients. Not only for Indonesian community, but also worldwide.

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The absence of genetic abnormalities in the coding region of the genes that was screened indicates that genes tested in this study in this population have very low frequency. Nevertheless, new unclassified variants in the patients list were found. Results also strengthen the importance of genetic counselling especially parental testing before conclude any mutation finding on sequencing method.

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