

Establishing Fragile X Research Center: An Experience from Developing Country, Indonesia

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Abstract

Fragile X syndrome (FXS) is the most common cause of monogenic inherited intellectual disabilities (ID) with X-linked pattern of inheritance. The prevalence of ID in developing countries was calculated as much as 3%, of which genetic was the underlying mechanism in about 20% of the cases. In Indonesia, fragile X syndrome accounted for 2% of the ID population. Center for Biomedical Research (CEBIOR), Faculty of Medicine Diponegoro University is the only Fragile X Research Center in Indonesia located in Semarang, Central Java, is the only center that offered FXS molecular diagnosis. Based on the number of the Indonesian population, 28,000 cases of FXS should be diagnosed in our center. However, the phenotype of FXS is less obvious when compared to Down syndrome (the most common cause of genetic ID) resulting in indistinguishable FXS. Coupled with the fact that the knowledge of FXS in healthcare providers concerning several aspects of FXS is yet to be fully understood, hence the awareness of FXS is unsatisfactory. Another obstacle that may have caused low detection rate is due to the lack of attention in genetic disorders from the government. In Indonesia, infectious disease is still the most important issue for national insurance health coverage followed by metabolic syndrome and cancer which may yield a direct impact on the mortality rate. FXS screening program based on research in high risk ID population had been done using various techniques, ranging from conventional cytogenetics to advanced molecular analysis in our center. Concerted effort is needed to improve the general awareness of the syndrome and encouragement from the government may shift national insurance health coverage in genetic disorders to be more favorable.

Keywords: Fragile X syndrome; Indonesia

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