

Blood Spots Screening for the Identification of Expanded Alleles in the *FMR1* gene among Intellectual Disability Students in Indonesia using Rapid Polymerase Chain Reaction

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Fragile X Syndrome (FXS) is the most common inheritance cause of intellectual disability (ID) and autism. The prevalence of full mutation was estimated to be 1 in 2633 and the premutation 1 in 251. Screening in Indonesia showed that FXS prevalence is 1.9%. The advances of targeted treatments in FXS have led to a newborn and high risk populations of FXS. Tassone (2008) introduced a rapid and inexpensive method for screening *FMR1* allele sizes throughout the pre-mutation and full-mutation range using a dried blood spot which suitable for screening large populations.

We involved 107 students (67 males and 50 females), ranging from 5-23 years of age from special school for ID in Semarang, Central Java, Indonesia. Our preliminary data indicates the presence of one male with an expanded allele in this study. Confirmation for the presence of a full mutation allele and the determination of the methylation status in this subjects by using Southern Blot analysis (standard method), is still in progress.

As conclusion, blood spots PCR screening is an effective method to conduct a test in high risk population especially in the remote area far from laboratory facilities. We are increasing the number of sample to determine the prevalence of FXS in this population and going to implement the treatment for individual with FXS.

Keywords: screening, *FMR1* gene, fragile X syndrome, rapid polymerase chain reaction