Hair root FMRP expression for screening of fragile X full mutation females

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First Author	
Last Author	
Authors	Rujito, L; Kustiani, D; Severijnen, LA; Hanzon, P; Faradz, SMH; Willemsen, R;
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Abstract	The fragile X syndrome is the most common form of inherited mental retardation in humans, caused by an expansion of the cytosine-guanine-guanine (CGG) repeat in the fragile X mental retardation 1 (FMR1) gene located on the X chromosome. Antibody tests have been developed to identify fragile X patients, based on the presence or absence of fragile mental retardation protein (FMRP) in both lymphocytes and hair roots. The objective of this study was to compare correlations of hair root and lymphocyte FMRP expression with cognitive functioning in female rural area probands carrying the full mutation. Thirty females (normal, premutation, or full mutation) were selected from Indonesian fragile X families and were tested for FMRP expression in lymphocytes and hair roots using the FMRP antibody test. Subject genotype was determined by Southern blot analysis, and IQ equivalent by Raven's Standard Progressive Matrices. Statistical analysis was by Pearson correlation. FMRP expression in blood lymphocytes was relatively higher than that in hair roots, but hair root FMRP expression was strongly correlated with cognitive functioning in female full mutation carriers (r=0.64, p=0.015), whereas no significant correlation between lymphocyte FMRP and cognitive functioning was found (r=0.31, p=0.281). Around 14% of subjects had a normal and 7% a borderline IQ level, while 79% had mild mental impairment. In conclusion, hair root FMRP expression may be a useful marker for identification of fragile X full mutation females.
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Author	Dr Dr LANTIP RUJITO, M.Si.Med