Title of dissertation				
Low prevalence of the BCR-ABL1 fusion gene in a normal population in southern				
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Background and Purpose: *BCR-ABL1* fusion gene is the driver mutation of Philadelphia chromosome-positive chronic myeloid leukemia (CML). Its expression level in CML patients is monitored by using real-time quantitative polymerase chain reaction defined by International Scale (qPCR^{IS}), in order to guide disease treatment. The *BCR-ABL1* has also been found in asymptomatic normal subjects. Previous studies performed convenient sampling and non-qPCR^{IS} method to study normal subjects harbouring *BCR-ABL1*, thus those results could not be inferred to the prevalence in normal population. We have conducted a normal population study in order to determine the prevalence in normal population harbouring *BCR-ABL1* using qPCR^{IS}.

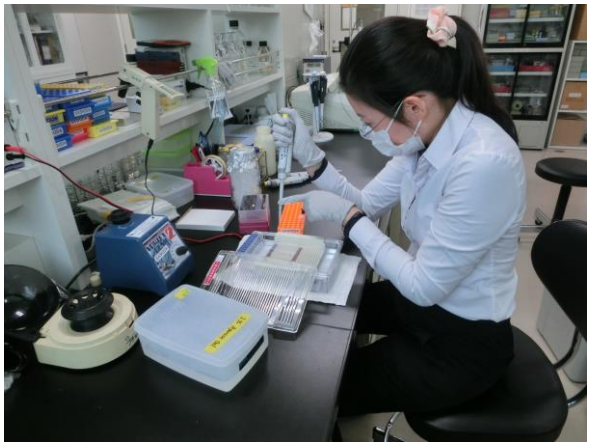
Methods: This was a cross sectional community-based study studying the southern adult Sarawak population using a two-stage sampling method based on Malaysia Department of Statistics population survey procedure. The sampling frame was set at two divisions in southern Sarawak and was divided into enumeration block (EB) and subdivided into living quarter (LQ). The first stage stratified sampling selected the EBs based on the population density of the two divisions. The second stage cluster sampling selected 12 LQs out of all the LQs in each EB. All eligible subjects within a LQ were recruited. qPCR^{IS} was performed using a validated commercial kit. Both *BCR-ABL1* and control *ABL1* gene were amplified in two replicates for each sample.

Results: A total of eight EBs, 88 LQs and 190 subjects were studied and analysed. 146/190 (76.8%) and 102 (53.7%) had satisfying sum of *ABL1* >20,000 and >100,000 copy number, respectively. We found one positive subject showing 0.0023%^{IS} with sum of *ABL1* 126,575, and repeated qPCR^{IS} of this subject was 0.0032%^{IS}. Sanger sequencing of this amplified product confirmed *BCR-ABL1* e13a2 transcript.

Conclusions: We herein demonstrated that the *BCR-ABL1* fusion gene is expected to be present in approximately 0.5–1% of normal adult individuals in southern Sarawak.

Photos

Meeting (for thesis defense)



Experiment