**Cytogenetic Analysis for Research and Services**

**Sultana MH Faradz**

Center for Biomedical Research, Faculty of Medicine, Diponegoro University

**Abstract**

That the correct chromosome number in man is 46 was first recognized by Tjio and Levan in 1956. Perhaps few Indonesians know that Tjio was an Indonesian scientist studying in Sweden and then living in the US. Cytogenetic analyses are commonly performed to determine both structural and numerical chromosome aberration, whilst changes in chromosomes can lead to birth defects, syndromes, or even cancer. Several chromosomal aneuploidy syndromes were identified after the establishment of various chromosome banding techniques in late 1960’s. Specific cell culture media was found to express fragile site in the beginning of 1970’s and since then, inherited Fragile X Mental Retardation syndrome could be diagnosed. However, some female permutation cases have been often misdiagnosed. Further molecular analysis has resolved this problem by revealing more CGG repeats in the promoter region *FMR1* gene, which is related to the expression of fragile site and the severity of the diseases.

In Disorder of Sex Development (DSD), early gender assignment and reconstruction surgery has been challenged because of the dilemma of gender identity development in later life. Cytogenetic analysis for the first-line gender assignment is important in newborn with DSD. Proper diagnosis with hormonal and mutation analysis should be elucidated to avoid medical, psychological, and social aspect in adult life. The most frequent genetic cases in our clinical experiences have been Androgen Insensitivity Syndrome and Congenital Adrenal Hyperplasia. Female Complete Androgen Insensitivity Syndrome (CAIS) with main symptom primary amenorrhea without cytogenetic analysis has often been diagnosed as inguinal hernia because of testicle location and size.

Diagnosis and treatment of several leukemias and lymphomas, as well as some solid tumors, depend on cytogenetic analyses to demonstrate consistent, specific chromosomal aberrations. Chromosome analysis in hematologic malignancy is indicated to support diagnosis, select therapy regimen, and elaborate prognosis. Specific chromosome translocations have been identified for hematologic malignancy. The breakpoints of several of these translocations have been cloned. Several loci of oncogene have been identified and sequenced. Molecular genetic analysis will replace cytogenetic analysis and shift the requirement for studying metaphase cells. Therefore, chromosome analysis in genetic disease and cancer should be attained with advanced molecular techniques, such as Fluorescence In Situ Hybridization (FISH) and microarray CGH analysis. Cytogenetic analysis is still useful and applicable in genetic disease diagnosis, sexual assignment, and hematologic malignancy in the laboratory with minimal equipments. Molecular analysis as a part of health care services in Indonesia has been limited in research centers in university setting; therefore, a comprehensive diagnosis with genetic analysis has often been improbable.

***Keywords***: cytogenetics, chromosome, syndrome, DSD, hematologic malignancy

Email: [sultanafaradz@gmail.com](mailto:sultanafaradz@gmail.com)