

ABSTRACT

Background and aim : Turner's syndrome is one of the most common chromosome abnormalities in female, with the prevalence of 1 in 2000 female live births. The main complaints are short stature and primary amenorrhea or infertility which gives rise to poor self esteem. The purpose of this study are : to get scientific information concerning chromosomal regions responsible for clinical features of Turner's syndrome especially short stature and primary amenorrhea, as well as the variation of cytogenetic abnormalities and its percentage among population of Turner Syndrome patient which were referred to Clinical Genetic Unit of Airlangga University Medical School/Dr. Soetomo Hospital.

Method : This research was descriptive study. The sample derived from Turner's syndrome patients which were referred to Clinical Genetic Unit of Airlangga University Medical School/Dr. Soetomo Hospital with the main complaint of short stature and primary amenorrhea with the age of patient 16 years old or older.

Results : The cytogenetic abnormalities of 46 patients are as follows, 30 individuals (65,21%) with the karyotype 45,X; 8 individuals (17,39%) with 45,X / 46,XX; 3 individuals (6,53%) with 46,X,i(X)(q10); 2 individuals (4,35%) with 46,X,del(X)(q10); 1 individuals (2,17%) with 46,X,del(X)(p10); and 2 individuals (4,35%) with a very rare case of 46,X,ter rea (X;X).

Conclusion : there are several differences in the variation of chromosome abnormalities and its percentage from that of previous studies of caucasian population and there is putative chromosome region distal to Xp22.3 that could be responsible for clinical feature of Turner's syndrome, namely primary amenorrhea, and there is 25% correlation between cytogenetic abnormalities and short stature.

Keywords : Turner's syndrome, cytogenetic (chromosomal) abnormalities, chromosomal region