Abstract

Turner syndrome (TS) is a genetic anomaly occurring in women with worldwide frequency 1:2,000. Turner syndrome's possible causes include X chromosome monosomy, mosaic karyotype (45,X/46,XX; 45,X/46,XY; 45,X/47,XXX), X isochromosome (46,X,i(Xq)), ring X chromosome (45,X/46,X,r(X)), and other X chromosome aberrations (deletion and translocation). Patients with Turner syndrome (with the exception of ring chromosome abnormalities) aren't diagnosed with mental retardation. Turner syndrome is closely related to the regulation of gene expression level, particularly with X chromosome inactivation. Genes escaping the X chromosome inactivation process in 45,X women are expressed in half the dose of a healthy woman. An example of such a gene is *SHOX*, which I decided to focuse on in this thesis. SHOX gene haploinsufficiency causes major Turner syndrome phenotype manifesting of short body and bone abnormalities. The research of other genes with possible roles in Turner syndrome is complicated by the absence of adequate model organism, which could be used for TS study with possibility to extrapolate the results to humans. In mice, both the inactivation process itself is different and the phenotypic manifestation of X monosomy (39, X) is also much milder than in 45,X women. This difference could be explained by the fact that in mice only about 3 % of genes escape inactivation, while in humans 15 % of genes escape.

Key words: Turner syndrome, Turner syndrome karyotype, X chromosome inactivation, genes escaping the X chromosome inactivation process, *SHOX* gene