Summary

The occurrence of ocular disorders in patients with Turner syndrome

Turner syndrome is among the most common chromosomal aberrations. It is caused by a missing or structural anomaly of one X chromosome, alternatively a chromosomal mosaicism. It is often connected with a more frequent occurrence of some ocular diseases. In our study 81 girls and women with Turner syndrome from the age of 5 to 23 years old were repeatedly examined. The occurrence of ocular diseases and their possible connection with karyotype was the main focus of our attention. Myopia (29 %) had the highest incidence in these girls, further there were hyperopia (24 %), epicanthus (20 %), colour vision deficiency (17 %), amblyopia (12 %), strabismus (10 %) and ptosis (5 %). Colour vision deficiency was defined as protanopia in 8.5 %, deuteranopia in 3.4 % and tritanopia in 5.2 %. The occurrence of colour vision deficiency was higher than in the general population where it differs in sexes. It is more often found in men. It is connected with the gene location for the red and green photo pigment which is localized on the long arm of the X chromosome. The occurrence of strabismus and ptosis was higher than in the general population. The total range of refractive errors was slightly higher than in the general population, with a different distribution according to karyotype. Hyperopia was recorded more often at the 45, X karyotype, namely 28 %, while for chromosomal mosaicism it was only in 18%. For myopia the ratio was reversed – chromosomal mosaicism in 31 % and in 45, X karyotype in 26 %. In total, while comparing individual eye defects incidence in 45, X karyotype and chromosomal mosaicism, similar findings were recorded. These results were also assessed with the help of statistics. At the standard 95% confidence level, no statistically significant relationship has been found between karyotype and any type of ocular diseases. Therefore, it can be concluded that at least within the studied sample group, karyotype has no influence on the occurrence of any specific eye defect.