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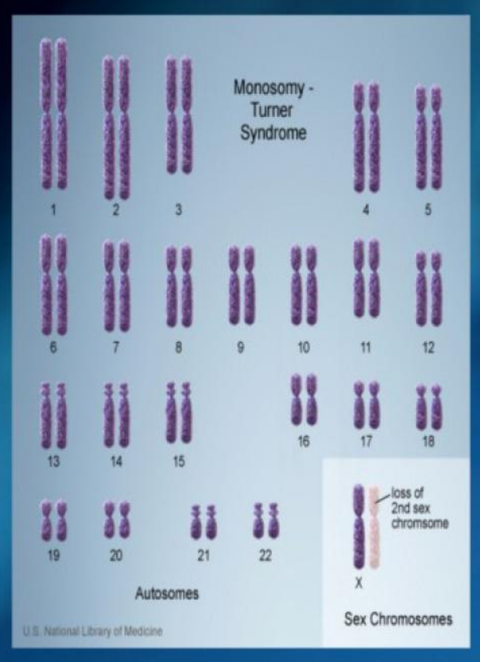

Results of molecular genetic studies for determination of latent mosaicism and parental origin of X chromosome in girls with Turner syndrome in Uzbek population

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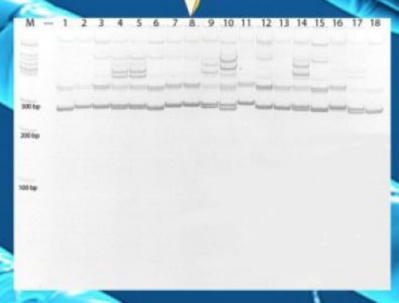
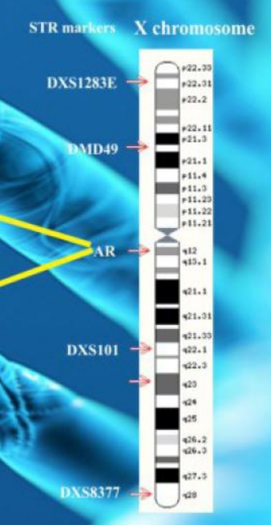
Turner Syndrome

Turner syndrome(TS) is one of the most common chromosomal abnormality syndromes, affecting 1 in 2,500 live born females in Uzbekistan

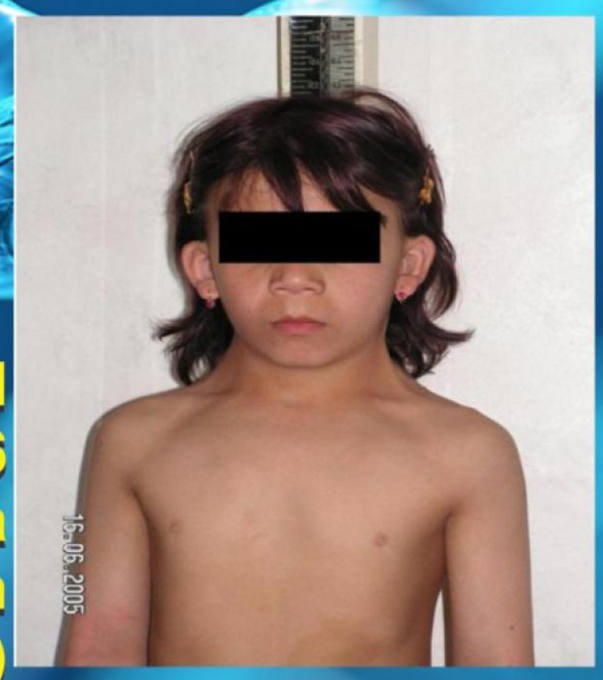
Goal: Identification of latent mosaicism and determination of a parental origin of an X chromosome in TS patients in Uzbek population.





Materials & Methods:

Molecular genetic studies are carried out in 35 patients with TS (26 with monosomy, 9 with mosaicism) at the age of 7 to 16 and their parents with a set of DIATOMTMDNA prep 200reagents. DNA amplification was performed in Applied Biosystemsthermocyclers. PCR products were subjected to electrophoresis on 12% acrylamide bis-acrylamidegel (29:1) with subsequent DNA staining with ethidium bromide and visualization by a BioDocAnalyze (Biometra) system.



Results:

Three X-linked markers (DMD 49, AR and DX1283E) were studied on the basis of their high level of heterozygosis (varying from 88.6 to 93.3%), a number of alleles (11 to 19) and localization both on a short and long X chromosome arm. The results obtained confirm that the use of a set of these primers (DMD 49, AR and DX1283E) will allow enhancing a probability of obtaining an informative marker and detection of latent X-mosaicism. Monozygosity on all 3 markers indicates the presence of only one X chromosome that in female patients will correspond to true monosomy (X0). Heterozygosis of one marker suggests on the presence of an additional second X chromosome or a part of an X chromosome which is observed both in 46XX karyotype (healthy) and in mosaic variants of chromosomal anomalies (45X0-46XX, 45X0-46XY).

Conclusion

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A comparative analysis of polymorphic markers in TS patients and their parents enable us to establish the origin of an X chromosome and determine in gametogenesis of which parents meiotic impairment occurred. Identification of mosaicism in Turner syndrome is very important from the viewpoint of setting correlations between a phenotype and karyotype.

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