AB072. Double aneuploidy of Down-Turner syndrome and Down-Klinefelter syndrome: case report and review

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**Background:** Down syndrome is the most common chromosomal abnormality in humans with an incidence of 1 in 770 live births. Chromosomal aneuploidy is quite frequent and may involve autosomes, as in Down’s syndrome, or sex chromosomes. Sex chromosomal aneuploidy Down-Klinefelter and Down-Turner syndrome are very rare. Objective of the study was to describe two cases of double aneuploidy: Down-Turner and Down-Klinefelter syndrome.

**Methods:** Sterile peripheral blood specimens with heparin for anti-coagulation were cultured in the RPMI medium in 72 h, harvested and performed the G-banding. The metaphase chromosomes were captured by the Karl Zeiss microscope system, analyzed by using Ikaros software (Metasystem) following the ISCN guidelines.

**Results:** Case 1 was a 9-year-old female patient with Down syndrome facie, short neck, developmental delay, mental retardation, and karyotype of 47,X,i(X)(q10),+21. Case 2 was a 2-month-old male infant with clinical signs as follow: Down syndrome facie, short neck, developmental delay, mental retardation, karyotype of 48,XXY,+21, and nondemonstrable testes on inguinal ultrasonography.

**Conclusions:** Down-Turner syndrome and Down-Klinefelter syndrome are rare conditions. Karyotyping should be performed for all patients with suspected Down syndrome regardless of the young age of the parents.

**Keywords:** Down-Turner syndrome; Down-Klinefelter syndrome; double aneuploidy

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