

Complex Genetic Disorders, Genetic Susceptibility to Infections

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

Moc Thi Thanh Hoang, Nhung Thi Hong Dinh, Nga Thi Tran, Lan Thuy An, Quang Dinh Vu, Huy Xuan Nguyen, Ngoc Diem Ngo, Hai Thanh Le

Human Genetics Department, Children's Hospital, Hanoi, Vietnam

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

doi: 10.21037/atm.2017.s072

Cite this abstract as: Hoang MT, Dinh NT, Tran NT, An LT, Vu QD, Nguyen HX, Ngo ND, Le HT. Double aneuploidy of Down-Turner syndrome and Down-Klinefelter syndrome: case report and review. *Ann Transl Med* 2017;5(Suppl 2):AB072. doi: 10.21037/atm.2017.s072