Abstract:
Objective: Isodicentrics (idic) are one the most cytogenetically common abnormalities of chromosome Y. The association of it with male non-obstructive infertility has been proven. Etiologically, it has wide spectrums of manifestations, ranging from females with Turner symptoms to males with spermatogenic failure. The central aspect of this retrospective study is to report on nineteen infertile patients who had a de novo form of same idic(Yq) karyotype with variable degrees of mosaicism.
Cases: The probands were nineteen infertile patients including: eighteen male and one female patient who suffered from infertility with same idic(Y) karyotype and referred to our center. Intervention(s): Cytogenetic methods on blood samples, fluorescence in situ hybridization (FISH) on seminal germ cells and blood, and PCR-based molecular approaches were carried out.
Result(s): The results obtained from cytogenetic analysis revealed abnormal Y chromosome: 45,X/46,X,idic(Y)(q11.22). The investigation followed by FISH technique which confirmed a rearranged Y chromosome, with two centromeres and two SRY signals, plus a cryptic marker chromosome with various levels of mosaicism. The aneuploidy of sex chromosomes was also detected in haploid seminal germ cells. Multiplex PCR analysis of blood samples demonstrated microdeletion in AZFb and AZFc loci.
Conclusion(s): By virtue of structural similarity between inv(Y) and idics(Y), usage of confirmatory techniques, e.g. FISH or PCR-based methods, could help avoid errors in cytogenetic results, and precisely delineate chromosomal aberrations in patients with fertility difficulty when clinical data fails to clarify the cause of infertility.