عنوان مقاله:

Cytogenetic and molecular genetic analysis of dicentric Y chromosome and its relation to male azoospermia

محل انتشار:

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خلاصه مقاله:

Background: Cytogenetic analysis, Y-chromosome microdeletion screening, FISH techniques and other genetic methods have helped in finding the causes of infertility in azoospermic or severe oligoazoospermic cases in the last decade. Objective: In the present study, we characterized an abnormal Y-chromosome, detectedas a mosaic in an azoospermic male ascertained for infertility. Materials and Methods: Chromosome analysis, using G, Q and C banding techniquesand FISH analyses with several different DNA probes specific for Y and X chromosome sequences [XY centromeric α-satellite, Y non-α-satellite III, LSI-probes ofthe Y chromosome, WCP of Chromosome Y, SRY gene, subtelomeric Xp and Yp, which cover the SHOX (short stature-homeobox containing) gene, and subtelomeric Xq and Yq probes] were performed. A total of 20 sequence tagged sites were analyzedusing primer sets specific for the Y-chromosome microdeletion loci. The primers were chosen to cover AZFa, AZFb, and AZFc regions as well as the SRY gene.Results: Chromosome analysis revealed a gonosomal mosaicism of monosomy X (51%) and a pseudodicentric Y (49%) chromosome: mos 45, X/46,X psu dic (Y)(qter→p11.32 :: p11.2→qter). Molecular genetic studies did not show deletions in the AZFabc regions, but a deletionwas found in the short arms of the dicentric Y chromosome. One of the SRY genes was also missing. Conclusion: The azoospermia in this patient could be explained by either the presenceof an abnormal Y-chromosome that cannot form a sex vesicle (which appears to be necessary for the completion of meiosis process and the formation of sperm), or the presence of the 45, X cell line.

کلمات کلیدی:

Dicentric Y chromosome, Dic (Yq), FISH, Mosaicism, Azoospermia