Chapter VI

Correlation of CA, Yq microdeletion M470V polymorphism and male infertility
6.1. Introduction

Studies have reported that environmental and genetic factors are believed to be responsible for a marked decline in male reproductive health and an increase in the population of infertile men. Increasingly, genetic factors are recognized and their identification is essential in informing couples about the prospect for pregnancy outcome, transmission of infertility and/or non-gonadal disease in offspring. The CA are most common cause that results in sterility or degrees of infertility. Severely impaired sperm production is associated with a significantly higher frequency of both numerical and structural chromosomal abnormalities [1]. In addition to major CA, Y-chromosome microdeletions and multiple gene variants controlling the male fertility like CFTR gene polymorphisms are shown to be the causative factor, all of which need special consideration. As the study analyzed the CA, Yq microdeletion (AZF) and CFTR polymorphism, in infertile men, an attempt was made to evaluate the influence of all those three parameters on male factor infertility.

Figure 6.1. Correlation of CA, Yq and CFTR gene M470V polymorphism in infertile men
6.2. Results and discussion

This study was conducted to assess the prevalence and risk of transmitting genetic abnormalities; the results of combined andrological, cytogenetic and molecular genetic screening for Yq microdeletion and *CFTR* gene polymorphism in \(n=175\) infertile men who were candidates for assisted reproductive technology is depicted in Figure 6.1. The obtained results showed that among infertile men, (i) 3.57% had both CA and Yq microdeletion; (ii) 3.89% had CA and MV heterozygotic alleles and (iii) 15.05% had Yq microdeletion, and MV heterozygosis in *CFTR* gene. Only 2.02% cases showed CA, Yq microdeletion and M470V polymorphism. A substantial number of infertile men; however, present with a history associated with fertility problems had normal findings on genetic testing. However, the availability of novel whole genome approaches encourage future studies, and will likely contribute to the identification of recurrent genetic factors. Analysis of CA in meiotic chromosomes is an alternate method as an increased prevalence of aneuploidy in the ejaculated or testicular sperm of infertile men with a normal peripheral blood karyotype was also reported and may account for higher rate of *de novo* abnormalities in ICSI offspring [2].

References
