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Analysis of the AZF Region in Slovak Men with Azoospermia

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Abstract : Y chromosome microdeletions are the most common genetic cause of male infertility and screening for these microdeletions in azoospermic or severely oligospermic men is now standard practice. Analysis of the Y chromosome in men with azoospermia or severe oligozoospermia has resulted in the identification of three regions in the euchromatic part of the long arm of the human Y chromosome (Yq11) that are frequently deleted in men with otherwise unexplained spermatogenic failure. PCR analysis of microdeletions in the AZFa, AZFb and AZFc regions of the human Y chromosome is an important screening tool. The aim of this study was to analyse the type of microdeletions in men with fertility disorders in Slovakia. We evaluated 227 patients with azoospermia and with normal karyotype. All patient samples were analyzed cytogenetically. For PCR amplification of sequence-tagged sites (STS) of the AZFa, AZFb and AZFc regions of the Y chromosome was used Devyser AZF set. Fluorescently labeled primers for all markers in one multiplex PCR reaction were used and for automated visualization and identification of the STS markers we used genetic analyzer ABi 3500xl (Life Technologies). We reported 13 cases of deletions in the AZF region 5,73%. Particular types of deletions were recorded in each region AZFa,b,c .The presence of microdeletions in the AZFc region was the most frequent. The study confirmed that percentage of microdeletions in the AZF region is low in Slovak azoospermic patients, but important from a prognostic view.

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