MÜSBED 2014;4(Suppl. 1):S14

Autosomal Gene Defects Investigation of Male Infertility in Germ Cell Aplasia Cases

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Objective: Sertoli-cell-only syndrome (SCOs), also called germ cell aplasia, describes a condition of the testes in which only Sertoli cells line the seminiferous tubules and is diagnosed by testicular biopsy. SCOs is considered as irreversible infertility. SCOs, a histological diagnosis, consists of multifactorial reasons including Y microdeletions, Kleinefelter syndrome, cystic fibrosis gene mutations, XYY syndrome, cryptorchidism, radiation, cytotoxic drugs and viral infections. Although the etiology of the disease is currently unknown, it is believed that autosomal gene defects could lead to SCOs. The aim of this study was to detect autosomal genetic defects and to determine candidate genes in SCOs infertile men.

Methods: Single nucleotide polymorphism + comparative genomic hybridization microarray technology (SNP+CGH array) was performed on 39 infertile patients with SCOs. Array CGH compares the patient's genome against a reference genome and identifies uncover deletions, amplifications, ploidy abnormalities and loss of heterozygosity (LOH).

Results: We examined a link between defected spermatogenesis genes and infertility. Detected amplifications and deletions in several genes are namely, SHBG, COL1A1, HOXD9, SYCE1, EMX2, EMX2OS, CATSPER2 and loss of heterozygosity in several genes are namely SPATA gene family (SPATA18, SPATA17, SPATA16, SPATA12, SPATA4, SPATA2), TSSK gene family (TSSK3, TSSK4, TSSK6), DNALI1, DNAH5, DNAH11, SPAG16, SPAG8, DMRT1, DMRT2, FSHR, LHCGR, GNRHR, SPACA1, SPACA3, TSGA10, SMCP, KIT, TCTE3, TEX14, FGF8. Amplifications and deletions were detected on some of the genes who play a role in epigenetic changes.

Conclusion: Epigenetic genes (H19, KCNQ1, IGF2, CDKN1C) are expected to be linked with male infertility.

Key words: SCOs, infertility, array-CGH, autosomal genes