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Reproductive Genomics: From Gene expression profiling to molecular diagnostics

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Spermatogenesis is a highly complex process and many genes are involved in its regulation, but relatively few genes have been identified. To gain a comprehensive view of gene expression and of the regulatory mechanism involved in germ cell development, we used a mouse DNA chip to compare the gene expression profiles of mouse pachytene spermatocytes and round spermatids, which were isolated from 9-week ICR mice testes by velocity sedimentation under unit gravity (VSUG). We verified differentially expressed genes by semi-quantitative and real-Time RT-PCR, and found that the genes identified include; primarily chromosomal genes related to intracellular enzyme and nucleic acid binding proteins, and kinase and substrate genes that mediate cell cycle transition. This dynamic gene expression and localization during spermatogenesis may be critical for the meiotic cell division process and germ cell development. We also developed molecular diagnostic tools to monitor male infertility. The long arm of the human Y chromosome is required for male fertility. Deletion in three different regions can cause severe spermatogenic defects ranging from non-obstructive azoospermia to oligozoospermia. These molecular diagnostic tools provide economic and high-throughput methods for detecting the deletion of genomic DNA sequences of large groups of infertile patients, and a new approach to male infertility screening.

