

# Molecular Biology and Reproduction

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**Abstract.** Modern molecular biology has provided unique insights into the fundamental understanding of reproductive disorders and the detection of microorganisms. The remarkable advances in DNA diagnostics have been expedited by the development of polymerase chain reaction (PCR) and the ability to isolate DNA and RNA from many different sources such as blood, saliva, hair roots, microscopic slides, paraffin-embedded tissue sections, clinical swabs, and even cancellous bone. These technical advances have been bolstered by the development of an increasing number of effective screening techniques to scan genomic DNA for unknown point mutations. The continued development of technology will ultimately result in automated DNA (deoxyribonucleic acid) diagnosis for the practicing clinician. The continuing expansion of information concerning the human genome will place an increasing emphasis on bioinformatics and the use of computer software for analyzing DNA sequences. With the automation of DNA diagnosis and the use of small samples (500 nanograms), the direct examination of the DNA of a patient, fetus, or microorganism will emerge as a definitive means of establishing the presence of the specific genetic change that causes disease. A knowledge of the precise pathology at the molecular level has and will provide important insights into the biochemical basis for many human diseases. A firm knowledge of the DNA alterations in disease and expression patterns of specific genes will provide for more directed therapeutic strategies. The refinement of vector technology and nuclear transplantation techniques will provide the opportunity for directed gene therapy to the early human embryo. This presentation is designed to acquaint the reader with current techniques of testing at the DNA level, prototype mutations in the reproductive sciences, new concepts in the molecular mechanisms of disease that affect reproduction, and therapeutic opportunities for the future. It is hoped that future refinement of these techniques combined with the ability to maintain genetic modification of these cells with recombinant vector technology will provide a definitive therapy for many single gene disorders, such as sickle cell anemia and thalassaemia. It is truly the challenge of the next century to decipher how these legions of newly discovered genes work, and to create a molecular language that can extend across all organisms. (Keio J Med 48 (1): 12-21, March 1999)

**Key words:** molecular biology, fundamental techniques

## Techniques of DNA-RNA Analysis

Insights into the molecular pathology of reproductive disorders require a firm knowledge of the techniques to analyze DNA for the detection of mutants. The purpose of this segment is to describe those techniques of analysis with special reference to those gene systems that are relevant to the reproductive sciences. The role of RNA analysis will be discussed briefly with

future applications of these and other techniques to the diagnosis and study of reproductive disorders.

### *Polymerase chain reaction (PCR)*

Almost all of the diagnostic techniques discussed in this syllabus use genomic DNA isolated from nucleated cells in blood as the starting material. The next step after DNA isolation is the amplification of specific

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