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Edited by

Alan Thornhill, PhD, HCLD

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Preface

Until recently, the world of diagnostics revolved around large chunks of tissue, whole blood samples, cell suspensions, or extracted DNA in microgram quantities. Given such a large starting template, problems of contamination and assay failure are rare but do still occur. When working with extremely small templates such as single cells or small groups of cells and their metabolites, these diagnostic challenges are simply magnified. Single cell diagnostics is a fast moving area even though the field has focused primarily on the analysis of embryonic blastomeres for the preimplantation diagnosis of single gene and chromosomal disorders—a relatively tiny area of diagnostics in terms of test volume. In just over a decade, there has been a diagnostic shift in the field from relatively simple singleplex PCR assays analyzed with ethidium based gel electrophoresis through sensitive fluorescent PCR involving multiplexing to expression analysis of multiple genes. Finally, the seemingly ubiquitous microarray technology has been applied to single cells for the identification of chromosome abnormalities and DNA alterations. *Single Cell Diagnostics: Methods and Protocols* is intended for clinical and research scientists as well as those providing care for couples seeking treatment for infertility or preimplantation genetic diagnosis. The aim is for all readers to extend their knowledge and expertise in analysis of single cells (whether or not that is their specific need). The majority of readers may not require assays of such exquisite sensitivity, but it has been my experience that many excellent research and diagnostic laboratories have learned useful tips from those of us struggling to salvage accurate diagnostic information from a single cell without contamination. *Single Cell Diagnostics: Methods and Protocols* starts with laser-assisted cell collection, noninvasive assessment of single cells and moves through the techniques of standard fluorescence *in situ* hybridization and polymerase chain reaction (PCR). As the reader moves through the book, the scope and complexity of each technique gradually increases as real-time quantitative PCR, isothermal whole genome amplification, comparative genomic hybridization, real-time gene expression analysis and the production of RNA and cDNA libraries are covered. The book closes with the application of customized microarrays to the study of single cells.

The future may see (1) a further shift away from preimplantation genetic diagnosis and more toward more routine diagnostic analysis in diseases such

as cancer (in situations where very little tissue might be available for analysis) and (2) direct proteomic analysis and indirect analysis (via the secretome) from single cells.

Alan Thornhill, PhD, HCLD

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