Preimplantation Genetic Screening and Preimplantation Genetic Diagnosis

Chantae Sullivan-Pyke, MD, Anuja Dokras, MD, PhD*

KEYWORDS

• Preimplantation genetic screening (PGS) • Preimplantation genetic diagnosis (PGD)
• Comprehensive chromosome screening (CCS) • Next-generation sequencing (NGS)

KEY POINTS

- Preimplantation genetic screening (PGS) improves pregnancy rates by allowing the selection of euploid embryos for transfer. Euploid embryos are more likely to implant and develop into a healthy pregnancy.
- Preimplantation genetic diagnosis (PGD) facilitates the selection of unaffected embryos for transfer to the uterus to avoid transmission of disease-causing genetic mutations.
- Preimplantation embryos can be biopsied at either the cleavage stage or blastocyst stage of development. Blastocyst stage biopsy is advantageous compared with biopsy at the cleavage stage because more cells can be analyzed.
- Currently, comprehensive chromosome screening (CCS) platforms, such as array comparative genomic hybridization (aCGH), single nucleotide polymorphism (SNP) microarray, quantitative polymerase chain reaction (qPCR), and next-generation sequencing (NGS), are used for preimplantation genetic testing (PGT).

INTRODUCTION

Infertility affects 7.5 million women in the United States, and approximately 1 in 8 couples have difficulty conceiving or sustaining a pregnancy.[1] In vitro fertilization (IVF) is a successful treatment of infertility due to several causes, including tubal factor, male factor, and diminished ovarian reserve. Preimplantation genetic testing (PGT) can be performed on cells removed from early embryos before transfer to the uterus (**Fig. 1**)[2] for the purpose of preimplantation genetic diagnosis (PGD) or preimplantation genetic screening (PGS). PGD refers to the detection of known conditions, such as

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Division of Reproductive Endocrinology and Infertility, Department of Obstetrics and Gynecology, University of Pennsylvania, 3701 Market Street, Suite 800, Philadelphia, PA 19104, USA
* Corresponding author.
E-mail address: ADokras@obgyn.upenn.edu

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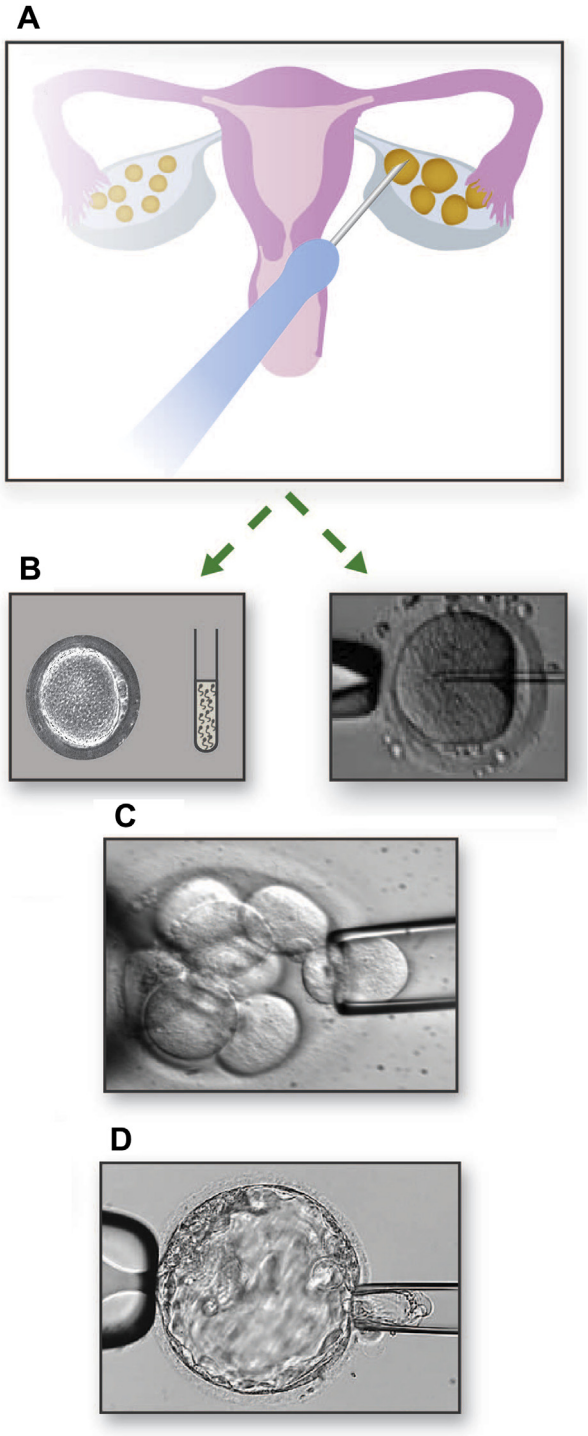

Fig. 1. IVF and embryo biopsy. (*A*) Cartoon showing the female reproductive system. Ovaries are stimulated by daily injections of gonadotropins, and oocytes are then retrieved under