

# Invitae preimplantation genetic testing

A critical step in optimizing IVF outcomes

Preimplantation genetic testing (PGT) increases the likelihood of IVF success by helping select the best embryos to:

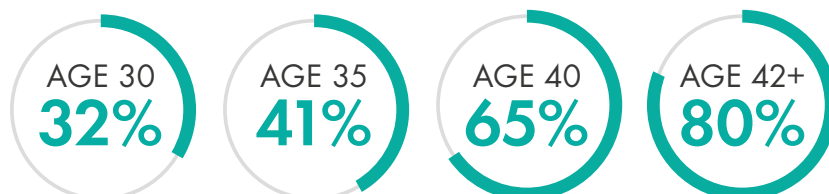
- Improve pregnancy rates<sup>1,2</sup>
- Lower miscarriage rates<sup>1,2</sup>
- Promote success in patients undergoing single-embryo transfer<sup>3</sup>
- Increase the likelihood of having a healthy baby<sup>2</sup>

IVF patients who may benefit from PGT include:

- Patients with a history of repeated implantation failure\*
- Patients with 1 or more miscarriages\*
- Patients who have had previous pregnancies with whole chromosomal abnormalities
- Patients with male factor infertility\*
- Women 35 years of age and older
- Patients interested in learning the sex of their embryo

## Maternal age and aneuploidy: Percentage of embryos with an abnormal number of chromosomes

Adapted from internal data on over  
90,000 embryos, 2018.



\*Invitae preimplantation genetic testing is designed to assess loss or gain of chromosomal material. It does not assess balanced changes within or between chromosomes.

### References:

1. Yang Z, Liu J, Collins GS, et al. *Mol Cytogenet.* 2012;5(1):24.
2. Keltz MD, Vega M, Sirota I, et al. *J Assist Reprod Genet.* 2013;30(10):1333-1339.
3. Forman EJ, Hong KH, Ferry KM, et al. *Fertil Steril.* 2013;100(1):100-107.
4. Gole J et al. *Fertil Steril.* 2016;105(2):e25.
5. Umbarger MA et al. *Fertil Steril.* 2016;106(3):e152.
6. Umbarger MA et al. *Fertil Steril.* 2017;108(3):e270.
7. Levy B et al. *Obstet Gynecol.* 2014;124(2 Pt 1):202-9.

## Invitae offers two types of PGT:

### PGT for chromosomal aneuploidy (PGT-A, also known as PGS)

- This test identifies embryos with the correct number of chromosomes to improve the chances of a successful pregnancy.
- When an embryo has extra or missing chromosomes, that's a condition known as aneuploidy. Most aneuploid embryos do not result in a healthy live birth: many lead to spontaneous miscarriage, some fail to implant in the uterus, and some result in a baby with birth defects, intellectual disability, and/or other health problems. Aneuploidy occurs across all age groups, but after a woman reaches age 35, the proportion of aneuploid embryos increases rapidly.
- Invitae PGT-A can detect the most frequent causes of miscarriage due to chromosome abnormalities.

Abnormality	Invitae PGT	Other NGS platforms	Frequency in miscarriages <sup>4-7</sup> (%)
Whole-chromosome aneuploidy	Yes	Yes	77.4–84.8%
Polyploidy (including 69,XXX)	Yes	Some	10.3%
Haploid/molar pregnancies	Yes	No	
Segmental aneuploidy	Yes	Yes	3.4%
Uniparental isodisomy (UPiD) *	Yes	No	<1%
Other	-	-	<1%

\*UPiD can be detected on all chromosomes except 17, 19, 20, 21, and 22.

### PGT for structural chromosome rearrangements/translocations (PGT-SR)

- This test evaluates embryos for unbalanced structural chromosome rearrangements (translocations). It is useful when a chromosomal translocation has previously been identified in the family.
- Invitae's PGT-SR also screens for chromosomal aneuploidy.
- PGT-SR cases are accepted on a case-by-case basis. A referral form and documentation of the known translocation (karyotype) must be submitted, reviewed, and approved by the Invitae Clinical Team to determine eligibility prior to following the steps below. The referral form is available at [www.invitae.com/forms](http://www.invitae.com/forms).

## How to order Invitae PGT

1. Request a collection kit at [www.invitae.com/request-a-kit](http://www.invitae.com/request-a-kit).
2. Ship frozen trophectoderm/blastocyst biopsy samples for overnight delivery, following the instructions included in the collection kit. The following is required for testing:
  - **Test requisition form (TRF):** Either a print or electronic TRF must be completed in order to proceed with testing. Download the paper TRF from [www.invitae.com/forms](http://www.invitae.com/forms) or talk to your client relationship manager to set up an online account to order electronically.
  - **Biopsy worksheet:** A completed biopsy worksheet form must accompany all samples.
3. Receive results in 5–7 days of sample receipt.

**Questions?** If you have questions about our offering or require technical support, we're here to help. Just visit [www.invitae.com/contact](http://www.invitae.com/contact).