Empowering informed choices that can change lives.

Genomic solutions in reproductive and genetic health.
Your committed partner across the care continuum.
Empowering informed reproductive choices. Improving inherited condition screening.

As a leader in innovative genomics technologies, Illumina is now your trusted partner for reliable answers along the reproductive and genetic health care continuum. Breakthrough products from both BlueGnome® and Verinata are now fully integrated into our portfolio. These additions expand our ability to serve the needs of clinical laboratories, healthcare professionals, and patients. Our industry-leading, next-generation sequencing (NGS), and microarray technologies deliver reliable, accurate information that can guide choices and transform lives.

Preimplantation Genetic Screening (PGS) and Preimplantation Genetic Diagnosis (PGD)

Improves in vitro fertilization (IVF) success rates by optimizing embryo selection. PGD can screen embryos when there is a risk of severe genetic disorders being inherited from parents.

Noninvasive Prenatal Testing (NIPT)

Delivers early, safe, and reliable answers about the chromosomal status of a developing fetus—minimizing procedure-related risks from invasive tests.

Cytogenetics

Identifies chromosomal aberrations responsible for genetic conditions that can cause developmental delay and/or congenital abnormalities.

Inherited Condition Screening

Helps detect carrier status or aid cystic fibrosis diagnosis or research the potential causes of undiagnosed inherited conditions.
Changing reproductive destinies by optimizing embryo selection.

PGS—the new standard of success in IVF.

While IVF has improved the treatment of infertility, its overall success rate remains low. Embryos with aneuploidy represent a major reason for IVF failure—causing problems with implantation or resulting in miscarriage within the first trimester. PGS is an effective method to screen for euploid embryos prior to implantation to significantly improve IVF success rates.

The PGS advantage
• Increased implantation rates—regardless of maternal age
• Lower risk of miscarriage
• Increased rates of ongoing pregnancy and live births
• Reduced chance of offspring with aneuploidy

What’s more, PGS can shorten the time needed to achieve success—reducing the number and cost of unproductive IVF cycles. PGS also empowers infertility experts to use single embryo transfers as a way of reducing the incidence of multiple births, which can be both complicated and costly, especially in women of advanced maternal age. With PGS, the overall result may be greater patient satisfaction and higher referral rates to IVF centers that routinely offer this option to their prospective parents.

Access a proven PGS portfolio
• 24sure Microarrays—Screen all 24 chromosomes to optimize selection of euploid embryos during IVF—and help improve success rates
• VeriSeq PGS Solution—Access our widely adopted NGS technology for comprehensive, accurate chromosomal euploid embryo screening and selection
• BlueFuse Multi analysis software—a multi-user database for storage, analysis, and reporting of results for PGS, PGD, and Cytogenetics

For Research Use Only
PGD—helping prevent serious gene disorders from being passed on.

PGD offers the powerful ability to screen for two important types of genetic defects. First, it can identify single-gene genetic conditions, such as cystic fibrosis (CF), sickle-cell anemia, and certain forms of muscular dystrophy, among others. This can help families avoid passing on a serious inherited disease to their offspring. PGD is also capable of screening embryos for chromosomal translocation disorders likely to cause miscarriage. This application may be especially valuable to couples with a history of miscarriage, which may be related to an inherited unbalanced translocation.

Our proprietary PGD screening solutions include:

- **karyomapping**—the most informative, genome-wide, single-cell test available, giving couples at risk of serious inherited diseases a new level of control over their reproductive destinies

- **24sure+™ Microarray**—screen for chromosomal translocations that can cause miscarriage—ensuring only embryos with no overall gain or loss of chromosome segments are transferred
The reassurance of knowing—simply, safely, sooner.

NIPT—reliable answers about fetal chromosomal status, without the risks of invasive procedures.

NIPT represents a major advance in prenatal testing, providing accurate information about fetal chromosomal status as early as 10 weeks gestation. It can detect the most common fetal chromosomal aneuploidies, with near-diagnostic accuracy, using a single maternal blood draw. In fact, NIPT is proven to be more accurate than traditional screening methods—even in a general obstetrical population.9

NIPT delivers reliable answers early and easily, providing parents with the peace of mind they need to make informed choices. NIPT also helps reduce the need for invasive procedures, such as amniocentesis or chorionic villus sampling (CVS). This is particularly meaningful for couples who’ve undergone fertility treatment.
The verifi® prenatal test is performed by Illumina in its Redwood City, California, CLIA-certified and CAP-accredited clinical testing laboratory. This test was developed by Illumina, and its performance characteristics were determined by Illumina. This test has not been cleared or approved by the U.S. Food and Drug Administration.
Timely insight into life-changing chromosomal abnormalities.

Cytogenetic testing—empowering deeper research into the causes of complex disorders.

Chromosomal abnormalities are a common cause of developmental delay and congenital conditions. These include syndromes, such as Down syndrome, DiGeorge syndrome, and Cri-Du-Chat syndrome, as well as other rarer microdeletion or microduplication disorders. Prenatal and postnatal cytogenetic research can allow for further insight into chromosomal aberrations.

Advanced cytogenetic microarrays—for a clearer view of genes.

Our industry-leading chromosomal microarrays include:

- **CytoChip™ Arrays**—Provide a greater understanding of the relevance and impact of chromosomal aberrations on complex diseases
- **Infinium® CytoSNP-850K Beadchip**—Offers comprehensive coverage of relevant dosage-sensitive genes for profiling chromosomal abnormalities—providing accurate cytogenetic data and capabilities that exceed standard cytogenetic analyses
  - Provides thorough coverage where you need it
  - Utilizes a wide variety of sample types, including blood, bone marrow, chorionic villi, and amniotic fluid
  - Delivers high-detection sensitivity

For Research Use Only
Identifying inherited conditions to make a difference for generations.

Detecting undiagnosed inherited conditions—for individual answers that affect an entire family.

Some inherited diseases may be immediately obvious at birth, while others may not be. Yet when such conditions run in a family, you want to be able to make a definitive diagnosis and provide reliable answers and peace of mind for everyone involved.

Many recessive, pediatric-onset Mendelian disorders are due to pathogenic mutations found in coding exons and intron-exon boundaries. NGS technology has been successful in finding causal variants of many such inherited and rare conditions.
Recognizing carriers of inherited conditions.

The ability to determine an individual’s carrier status for an inherited condition can be particularly empowering to a prospective parent concerned about passing on a genetic disorder. Carrier screening, in conjunction with genetic counseling of carriers, is an effective way to ensure genetic disease risk is detected early—preferably prior to conception. NGS expands carrier screening for mutations with potential links to genetic disease.11

Expanded carrier-screening capabilities

- Comprehensive coverage
- Improved detection rates
- The right answers the first time

We currently offer several NGS options to aid in the diagnosis of CF, determine carrier status for CF, and research other inherited conditions.

For *in vitro* diagnostic use only

- **MiSeqDx™ Cystic Fibrosis 139-Variant Assay**—The first FDA-cleared NGS assay that can be used to determine CF carrier status
- **MiSeqDx Cystic Fibrosis Clinical Sequencing Assay**—The first FDA-cleared assay that can be used as an aid in diagnosing CF
- **MiSeqDx Universal Kit 1.0**—An FDA-cleared sequencing solution enabling clinical labs to develop their own NGS assays for use on the MiSeqDx instrument

For research use only

- **TruSight™ Sequencing Panels**—Targeted NGS panels focused on specific genes or regions thought to be relevant to particular conditions, including recessive pediatric-onset diseases, autism, and inherited causes of cardiomyopathy

Together, our state-of-the-art assays provide highly accurate results that deliver a deeper understanding of genetic profiles related to inherited conditions.
Empowering informed choices in reproductive and genetic health.

- Trusted leader in innovative, accurate genomic technologies
- A comprehensive portfolio of genomic solutions to improve options in reproductive and genetic health
- Timely, reliable answers that empower more informed choices and enhance lives
- Your committed partner for generations to come

Learn more about our comprehensive portfolio of genomic solutions for reproductive and genetic health.

Visit www.illumina.com/rgh

References


A global genomics leader, Illumina provides complete sample-to-answer solutions to the research and clinical communities. Illumina technology generates 90% of the world’s sequencing data. Through collaborative innovation, we are fueling groundbreaking advancements in the fields of oncology, reproductive health, genetic disease, microbiology, agriculture, and forensic science.

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