

Preimplantation genetic screening solutions.



# Leading preimplantation genetic screening (PGS) solutions.

## Improve IVF success rates by optimizing embryo selection.

Illumina offers a full range of next-generation sequencing (NGS) and microarray assays designed to deliver fast, accurate information that can guide choices and transform lives.

While *in vitro* fertilization (IVF) has advanced the treatment of infertility, the process itself remains inefficient with low success rates.¹ Chromosome aneuploidy—an abnormal number of chromosomes—is believed to be the main cause of IVF failure, as most aneuploid embryos will not implant or will miscarry in the first trimester.² Aneuploidy increases considerably in embryos with increasing maternal age.³ Successful IVF procedures are dependant, in part, on successful selection of euploid—a normal number of chromsomes— embryos, which can be enabled using PGS.⁴

#### The advantages of PGS.

- Leads to greater implantation rates and improved IVF clinical outcomes<sup>5</sup>
- Mitigates several reproductive challenges associated with maternal age<sup>3</sup>
- Allows for single embryo transfers and higher patient satisfaction rates<sup>6,7</sup>

Illumina offers both sequencing and array technologies for all your PGS needs. The NGS-based VeriSeq™ PGS and 24sure® Microarrays confidently and accurately screen all 24 chromosomes in approximately 12 hours for optimal selection of euploid embryos.











## VeriSeq PGS Kit— The next-generation solution.

- Industry-leading data quality—90% of the world's sequencing data is generated using sequencing by synthesis (SBS) technology from Illumina
- Fast, streamlined workflow—Sample to answer in approximately 12 hours
- High-throughput analysis—Screen up to 24 samples per run
- Exceptional solutions—Easy workflows, accurate results, and high-throughput capabilities

#### Streamlined VeriSeq PGS Workflow

DNA	Library	> Sequencing	Data	Report
Amplification	Preparation		Analysis	Generation
Extract and amplify DNA using the SurePlex™ DNA Amplification Kit	Prepare libraries for sequencing on the MiSeq® System using the VeriSeq PGS Kit - MiSeq	Start MiSeq instrument  Add library to the ready-to-use flow cell	MiSeq System demultiplexes samples and aligns reads to the genome	Analyze data using BlueFuse® Multi analysis software Generate report

The VeriSeq PGS workflow provides a fast, end-to-end method for screening up to 24 samples in approximately 12 hours.



# Leading preimplantation genetic screening (PGS) solutions.

### 24sure Microarrays— Proven, accurate PGS results.

- Widely used, proven method—PGS method of choice for many of the world's reference laboratories; used to process over 350,000 samples
- Fast, reliable results Accurate assessment of all 24 chromosomes in approximately 12 hours
- Straightforward workflow—Streamlined protocols include minimal tube transfers, documented QC stages, and flexible stopping points

#### 24sure Laboratory Workflow

Amplification	Labeling	> Hybridization	Scanning	Data Analysis
Extract and amplify DNA using the SurePlex DNA Amplification Kit	Label amplified DNA using random primers and Cy dyes	Hybridize labeled DNA fragments to the 24sure microarray	Scan array using a laser scanner	Analyze data using BlueFuse Multi analysis software Generate report

The 24 sure laboratory workflow is a straightforward procedure that is completed in approximately 12 hours.









### Illumina solutions for PGS.

Product	Platform	Number of samples	Scalable	Turnaround time (24 samples)
VeriSeq PGS Kit	NGS	96 (4 x 24 sample runs)	Yes	12 hours from sample to result with 2.5 hours of hands-on time
24sure Microarrays	Arrays	24	No	12 hours from sample to result with 3.5 hours of hands-on time

## Get started with PGS today.

Product	Catalog Number
VeriSeq PGS Kit	
VeriSeq PGS Kit - MiSeq (96 samples)*	RH-101-1001
MiSeq Sequencing System	SY-410-1003
24sure Microarrays†	
24sure (v3.0) Microarray Pack (16 hybridizations)	PR-10-408702-PK
SurePlex DNA Amplification Kit	PR-40-415101-00

 $<sup>^{*}</sup>$  The VeriSeq PGS Kit - MiSeq includes the SurePlex DNA Amplification Kit, VeriSeq DNA Library Prep Kit-PGS, VeriSeq Index Kit-PGS, and MiSeq Reagent Kit v3-PGS.

 $<sup>\ \, \</sup>uparrow \, \text{Third-party array scanner needed for 24 sure \, Microarray \, workflow. \, Please \, contact \, your \, sales \, representative \, for \, more \, information.}$ 

# Leading preimplantation genetic diagnosis (PGD) solutions.

# Gain genome-wide insight into the inheritance of genetic disorders.

In preimplantation genetic diagnosis (PGD), embryos that do not carry a specific genetic condition are selected for transfer into the uterus to prevent couples from transmitting the condition to their child. Using genetic markers, it is possible to assess the likelihood of an embryo carrying a mutated version of a gene involved in a severe single-gene disorder or an unbalanced subchromosomal translocation that can lead to a miscarriage or an affected child.<sup>8,9</sup>

## Karyomapping—The most informative, genome-wide, single-cell PGD screen available.

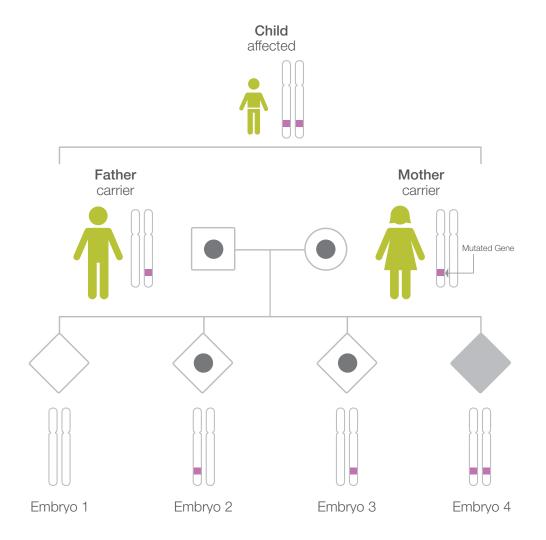


- Faster results—24-hour protocol eliminates the need for patientor disease-specific test development, significantly reducing the time from sample to answer
- Comprehensive coverage—Genome-wide, linkage-based analysis targets the most informative markers in the genome for insight into the inheritance of multiple single-gene disorders
- Accurate assessment—Karyomapping uses a large number of data points, reducing the risk of missing a recombination event



# Karyomapping can be performed on a wide range of recessive or dominant single-gene disorders.

Identifying the Inheritance Satus of an Embryo



In this illustration, parents of child with a recessive single-gene disorder are assessing the possibility of passing on the single-gene disorder to another child. Using karyomapping, the status of the embryo (affected, carrier, or unaffected) can be established.

# Leading preimplantation genetic diagnosis (PGD) solutions.

## 24sure+ Microarrays—Investigate subchromosomal imbalances in embryos.

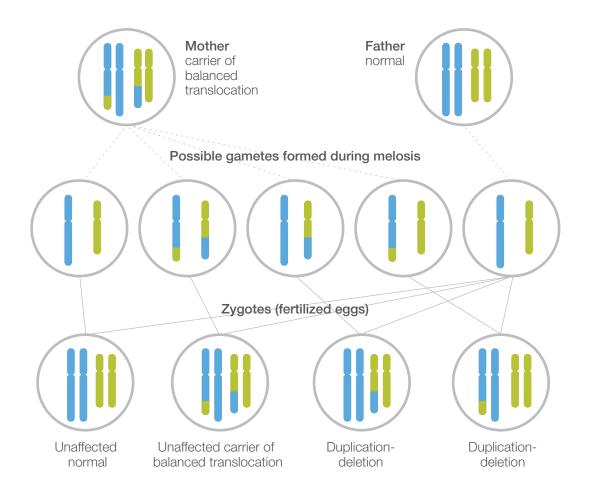
- Accurate identification—Increased coverage of pericentromeric and subtelomeric regions to ensure the reporting of the smallest derived chromosome fragments
- Fast, reliable results—Reporting in approximately 12 hours
- Straightforward workflow—Optimized protocols include minimal tube transfers, documented QC stages, and flexible stopping points





## 24sure+ Microarray investigates subchromosomal imbalances in embryos of reciprocal translocation carriers.

#### Inheritance of Unbalanced Translocations



Carriers of unbalanced translocations risk creating gametes with the rearrangements shown above. Using 24sure+ Microarrays, it is possible to identify embryos with chromosomal imbalances and ensure that only unaffected embryos are transferred in an IVF cycle.

# Leading preimplantation genetic diagnosis (PGD) solutions.

### Get started with proven PGD solutions today.

Product	Catalog Number
Karyomapping	
Infinium® HumanKaryomap-12 DNA Analysis (24 sample)	RH-103-1001
SureMDA™ DNA Amplification	PR-40-405102-00
iScan® System 110V/220V*	SY-101-1001
NextSeq® 550 Sequencing System*	SY-415-1002
24sure+ Microarray Pack <sup>†</sup>	
24sure+ Microarrays Pack (16 hybridization samples)	PR-10-408602-PK
SurePlex DNA Amplification kit (50 reactions)	PR-40-415101-00
SureRef Pack (16 reference hybridizations)	PR-40-415205-PK

<sup>\*</sup> The Infinium HumanKaryomap-12 BeadChip works with both the iScan System and the NextSeq 550 System. Only one system is needed.

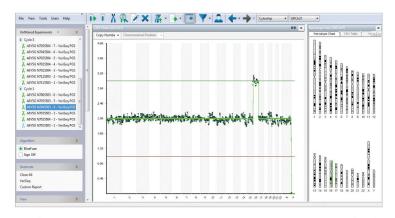


<sup>†</sup> Third-party array scanner needed for 24sure+ Microarray workflow. Please contact your sales representative for more information.



### BlueFuse Multi Analysis Software— Powering your PGS and PGD assays.

BlueFuse Multi software assists in the storage, analysis, and interpretation of all PGS and PGD data. All Illumina PGS and PGD kits include a software license for BlueFuse Multi Analysis Software.



#### User-Friendly BlueFuse Software Interface

BlueFuse Software ensures that laboratories can choose their preferred platform for any run without changing workflows. Generated data from one platform can be analyzed, stored, and viewed alongside data from the other. Data shown is for a euploid male embryo.



# World-class solutions. A community of support.



Illumina genomic solutions empower researchers and clinicians across the globe to find the answers they seek. When you join the Illumina community, you become part of a dynamic scientific movement that includes thousands of scientists, clinicians, and industry thought leaders. Throughout the year, we host user group meetings, symposia, consortia, online forums, and other initiatives—all designed to bring the best minds together to share ideas and advance science. In addition to on-site training, ongoing support, and phone consults, we offer webinars and courses at various Illumina locations. We're here with all the resources you need to advance reproductive and genetic health.

## Empowering breakthroughs in genetic screening.

A trusted leader in genomic solutions, Illumina now offers a comprehensive reproductive and genetic health portfolio. With breakthrough genetic screening, we are well-equipped to serve the needs of clinical research laboratories, healthcare professionals, and patients. Together, we can empower informed reproductive choices and improve the detection of genetic diseases.

Learn more about our reproductive and genetic health solutions.

Visit www.illumina.com/IVFProductBrochure

#### References

- 1. www.cdc.gov/art/artreports.htm
- Scott RT, Upham KM, Forman EJ, et al. Blastocyst biopsy with comprehensive chromosome screening and fresh embryo transfer significantly increases IVF implantation and delivery rates: a randomized controlled trial. Fertil Steril 2013;100(3):697-703.
- Harton GL, Munné S, Surrey M, et al. Diminished effect of maternal age on implantation after preimplantation genetic diagnosis with array comparative genetic hybridization. Fertil Steril. 2013;100(6):1695-1704.
- Fiorentino F, Bono S, Biricik A, et al. Application of next-generation sequencing technology for comprehensive aneuploidy screening of blastocysts in clinical preimplantation genetic screening cycles. *Hum Reprod*. 2014;29(12):2802-2813.
- Yang Z, Liu J, Collins GS, et al.
   Selection of single blastocysts for
   fresh transfer via standard morphology
   assessment alone and with array
   CGH for good prognosis IVF patients:
   Results from a randomized pilot study.
   Mol Cytogenet. 2012;5(1):24.
- Forman EJ, Hong KH, Ferry KM, et al. In vitro fertilization with single euploid blastocyst transfer: a randomized controlled trial. Fertil Steril. 2013;100(1):100-107.
- Grifo JA, Hodes-Wertz B, Lee HL, Amperloquio E, Clarke- Williams M, Adler A. Single thawed euploid embryo transfer improves IVF pregnancy, miscarriage, and multiple gestation outcomes and has similar implantation rates as egg donation. J Assist Reprod Genet. 2013;30(2):259-264.
- Fiorentino F, Spizzichino L, Bono S, et al. PGD for reciprocal and Robertsonian translocations using array comparative genomic hybridization. Hum Reprod. 2011;26(7):1925-1935.
- Ogilvie CM, Scriven PN. Meiotic outcomes in reciprocal translocation carriers ascertained in 3-day human embryos. Eur J Hum Genet. 2002;10(12):801-806.

**Illumina •** 1.800.809.4566 toll-free (US) • +1.858.202.4566 tel • techsupport@illumina.com • www.illumina.com

FOR RESEARCH USE ONLY

© 2015 Illumina, Inc. All rights reserved. 24sure, BlueFuse, Illumina, Infinium, iScan, MiSeq, SureMDA, SurePlex, VeriSeq, the pumpkin orange color, and the streaming bases design are trademarks of Illumina, Inc. and/or its affiliate(s) in the U.S. and/or other countries. All other names, logos, and other trademarks are the property of their respective owners. Pub. No. 1586-2015-001 Current as of 13 February 2015

