



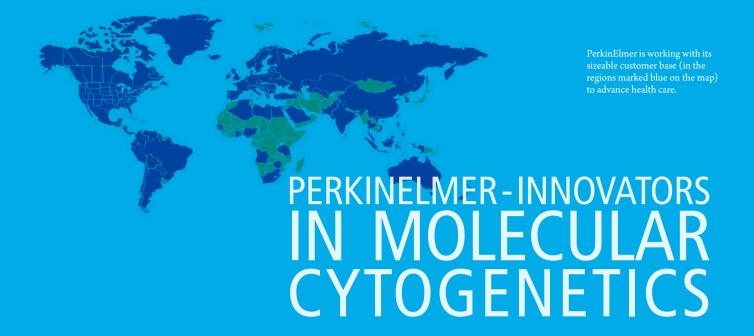


# A GLOBAL LEADER IN HUMAN AND ENVIRONMENTAL HEALTH

PerkinElmer is a global scientific leader committed to improving the health and safety of people and their environment. PerkinElmer is the world's leading supplier of newborn screening systems, a pioneer in methods for risk assessment during pregnancy and a leader in cord blood banking. PerkinElmer is also a pioneer in the field of molecular cytogenetic analysis continuously working towards innovation and development of cutting-edge technologies to improve human health. PerkinElmer's advanced chemagen technology provides cytogenetics and human genetic testing laboratories expertise in nucleic acid isolation across various sample types.

We aim to share our experience and expertise with the global cytogenetics community and significantly advance the field of cytogenetics.





#### Faster results – higher sensitivity

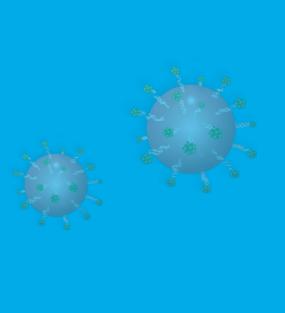
PerkinElmer is bringing the newest molecular techniques to cytogenetic laboratories.

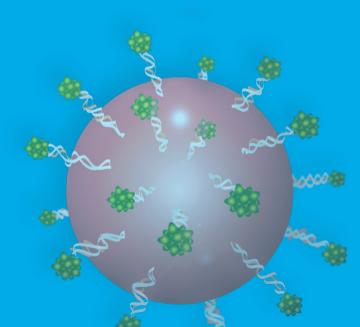
Our innovative methodologies support:

- Increased detection rates
- Faster results at lower costs
- Streamlined processes
- Support from sample to result

Oligonucleotide-based microarrays and bead-based multiplex BACs-on-Beads™ (BoBs) products have been specifically designed, developed and validated by cytogeneticists for cytogeneticists performing molecular karyotyping.

Together with chemagen, PerkinElmer is able to support the entire workflow incorporating automated DNA/RNA isolation solutions.







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## UNIQUE TECHNOLOGY FOR DNA/RNA ISOLATION

#### chemagen Technology – Complete Solutions for DNA/RNA Isolation in Human Research

A crucial step in human genetic research is the isolation of high quality DNA or RNA.

**chemagen Technology** offers flexible solutions for every application or daily throughput, special dedicated to the application fields listed:

- Human Genetics
- Biobanking
- Pathogen/Virus Detection

As the number of samples increases, the need for automation becomes more acute. Automation also offers better reproducibility and reliability in the isolation process. The key competence behind **chemagen Technology** is superior yields and high purity isolated genomic DNA and total RNA. This competence is expressed in chemagic Kit products, which contain proprietary magnetic particles with a high affinity to nucleic acids and low protein binding.

All products listed are for Research Use Only. Not for use in diagnostic procedures.



#### The chemagic 360 - outstanding flexibility

- Up to 4000 samples/day
- 1 96 samples / run
- 10 µl 10 ml sample volumes
- For blood, serum/plasma, saliva
- Compact design: 80 x 80 x 90 cm



#### The chemagic Prepito®-D -

#### innovative benchtop solution

- Up to 1 ml samples, 1 12 samples in parallel
- Pre-installed protocols
- · Cost-effective sample preparation
- · Automated precision for lower throughput



### chemagic Kits – magnetic beads for endless possibilities

- Specialized kits for isolation of genomic DNA and total RNA
- chemagen kits contain all reagents, components and plasticware



# CHEMAGIC KITS OVERVIEW

The chemagic Kit portfolio comprises a large amount of kits for the isolation of gDNA and total RNA.

Human Genetics and Biobanking	Instrument	Sample Volumes	Samples/Run
chemagic DNA Blood Kit special	chemagic 360	50 μl - 400 μl blood 1 ml - 4 ml blood 3 ml - 10 ml blood	96 24 12
chemagic DNA Blood Spot Kit special	chemagic 360	Paper filter punch outs	96
chemagic DNA Saliva Kit special	chemagic 360	4 ml saliva	24
chemagic DNA Buccal Swab Kit special	chemagic 360	1 swab	96
chemagic DNA Cell12M Kit special	chemagic 360	1.2 x 10 <sup>7</sup> cells	12
Prepito DNA Blood250 Kit	chemagic Prepito-D	100 μl - 250 μl	12
Prepito DNA Blood600 Kit	chemagic Prepito-D	600 µl	6
Prepito FFPE Kit	chemagic Prepito-D	10 µm section	12
Prepito DNA Tissue Kit	chemagic Prepito-D	animal tissues or cells	12
chemagic SEQ Pure Kit	Manual or on LH instruments	5 μl - 20 μl (without ethanol)	
Prenatal Genetics and Epigenetics	Instrument	Sample Volumes	Samples/Run
chemagic Circulating NA Kit Special	chemagic 360	1 ml serum/plasma 4 ml serum/plasma	12 24
chemagic Amniotic Fluid Kit special	chemagic 360	1 ml, 3ml amniotic fluid	12
chemagic Epigenetic NA Extraction Kit special	chemagic 360	5 ml plasma 4 ml plasma	12 12-24
chemagic Epigenetic Bisulfite Purification Kit special	chemagic 360	up to 500 µl bisulfite reaction mix	96
Prepito DNA Cyto Pure Kit	chemagic Prepito-D	250 µl blood, 10 mg tissue or 3-5 ml pelleted amniotic fluid	12
Prepito Circulating NA1k Kit	chemagic Prepito-D	1 ml serum/plasma	12
Isolation of total RNA	Instrument	Sample Volumes	Samples/Run
chemagic RNA Blood Kit special	chemagic 360	2.5 ml stabilized blood 2.5 ml stabilized blood	12 24
chemagic RNA Saliva Kit special	chemagic 360	1 ml serum/plasma	12
chemagic RNA Tissue Kit special	chemagic 360	50 mg tissue	12
chemagic mRNA/gDNA Kit special	chemagic 360	2 ml blood or bone marrow; max. cell number 20 x 10 <sup>6</sup>	12

Further chemagic Kits are available on request. Products listed are for Research Use Only. Not for use in diagnostic procedures.





## ARRAY CGH

### Oligonucleotide Array CGH Solution

Traditional karyotyping methods, such as FISH or G-banding, are well established for detecting chromosomal aberrations. However, these methods are limited by speed, throughput and resolution.

While these traditional methods are able to detect microscopic visible chromosomal alterations such as an extra chromosome band, smaller gains or losses in the genome cannot be found reliably. Array CGH in contrast enables higher resolution genome analysis and allows the detection of submicroscopic chromosomal imbalances across the entire genome in one single experiment.

CGX Oligo Arrays are Research Use Only products. Not for use in diagnostic procedures. CGX Oligo Arrays are manufactured by Agilent Technologies, Inc. PerkinElmer has developed an oligo array design for the detection of submicroscopic aberrations associated with learning disability and dysmorphic features. This array design allows researches access to relevant cytogenetic content through the use of our streamlined and robust CGX workflow.





## CGX OLIGO ARRAYS

## CGX Oligo Arrays are oligonucleotide-based microarrays

specifically designed for the detection of small genetic aberrations associated with

learning disability and dysmorphic features in research applications. The oligonucleotide probes on the CGX™, CGX™-HD and CGX™-SNP arrays cover the entire human genome with specific focus on over 245 cytogenetically relevant regions, 980 functionally significant genes, pericentromeric regions, and subtelomeres.

All three array types are based on the same design to provide the laboratory with throughput and detection flexibility. In addition, SNP probes on the  $CGX^{TM}$  -SNP arrays allow the detection of Absence of Heterozygosity (AOH) and Uniparental Disomy (UPD).

#### CGX OLIGO ARRAY CHARACTERISTICS THAT WILL BENEFIT YOUR WORK

- Proven design, representing over 245 known syndromic regions and 980 functionally significant genes
- Three different array types based on the same design to adjust for throughput and detection requirements

CGX Oligo Arrays are Research Use Only products. Not for use in diagnostic procedures. CGX Oligo Arrays are manufactured by Agilent Technologies, Inc.

CGX™	CGX™-HD	CGX™-SNP
8-plex array for detection of copy number abnormalities	4-plex array for detection of copy number abnormalities	4-plex array for detection of copy number abnormalities and Absence of Heterozygosity (AOH) and Uniparental Disomy (UPD)
Resolution of ~190 kb in the backbone, and ~28 kb in the targeted regions	• Resolution of ~40 kb in the backbone, and ~20 kb in the targeted regions	Resolution of ~80 kb in the backbone, and ~20 kb in the targeted regions
Evaluation of over 245 recognized genetic syndromes and over 980 gene regions of functional significance in human development	Evaluation of over 245 recognized genetic syndromes and over 980 gene regions of functional significance in human development	Evaluation of over 245 recognized genetic syndromes and over 980 gene regions of functional significance in human development
Covers subtelomeric and pericentromeric regions	Covers subtelomeric and pericentromeric regions	Covers subtelomeric and pericentromeric regions
		Detection of contiguous stretches of AOH (<10 Mb)
		Detection of triploidy



## PERKINELMER ARRAY SOFTWARE SOLUTION

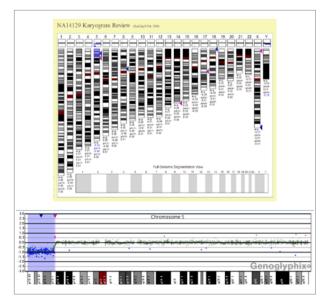
## Genoglyphix®: turning 55,000 samples into meaning

Genoglyphix® is a powerful data visualization software and database based on

the analysis of over 55,000 verified cytogenetic samples.

#### DESIGNED BY CYTOGENETICISTS FOR CYTOGENETICISTS

Developed by Cytogeneticists for Cytogeneticists, Genoglyphix® offers a proven analysis solution with a complete workflow including sample tracking, aberration categorization, final report creation functionality, and data sharing options with other centres. It also offers the possibility to create user-specific database tracks. Genoglyphix® provides intuitive data visualization and annotation features for streamlined, rapid and clear interpretation of CGX data.



 $Data\ visualization\ in\ Genoglyphix^{*}\ in\ Chromosome\ and\ Karyogram\ View$ 

## GENOGLYPHIX® – BRINGING MULTIPLE GENETIC DATABASES INTO ONE TOOL

- Access to Signature's database containing over 14,000 verified genetic alterations identified in over 55,000 samples
- Direct links to other relevant databases such as the DGV, OMIM, PubMed, UCSC and Ensembl for easy data interpretation
- Secure web based (128-bit encryption) access to Genoglyphix® database and software

## GENOGLYPHIX® EASILY GUIDES CYTOGENETICISTS THROUGH ANALYSIS WITH CONVENIENT TOOLS

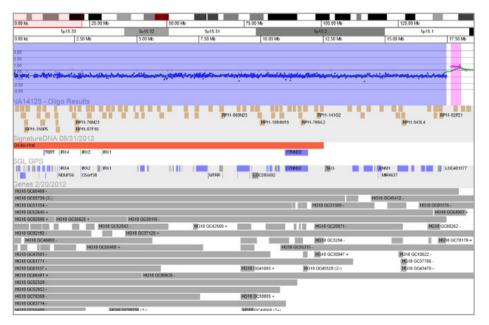
- Generation user defined databases and custom tracks displaying abnormal results, copy number variants and analysis notes
- · Report creation functionality
- Optional sharing of data with other Genoglyphix® users

Genoglyphix® software is for Research Use Only. Not for use in diagnostic procedures.

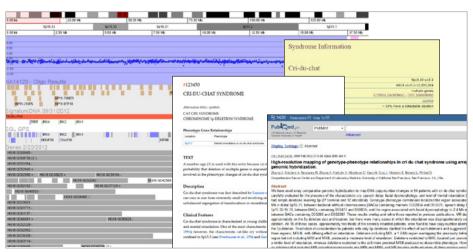


# GENOGLYPHIX® GENOGLYPHIX® BROWSER

#### GRAPHICAL REPRESENTATION OF ABERRATIONS IN THE GENOGLYPHIX® GENOME BROWSER



- < Syndrome track
- < Identification of relevant genes in the aberrant regions
- Identification of similar aberrations in Signature Genomics database of over 14,000 verified genetic alterations



Selection of the appropriate entries will automatically take you into databases such as OMIM, PubMed and Genoglyphix\* Syndrome Detail Page

Genoglyphix® software is for Research Use Only. Not for use in diagnostic procedures.



# CGXTHYBRIDIZATION CGXTHYBRIDIZATION

#### CGX<sup>™</sup> Hybridization Oven

The CGX™ Hybridization Oven is designed for optimal hybridization performance to achieve consistent and reliable results. The oven is a compact

and flexible instrument designed for an optimal microarray processing workflow.

#### • High Throughput

Designed to hold up to 24 CGX<sup>™</sup> Hybridization Chambers.

#### Consistent Data

Reproducible results.

#### Flexibility

- Variable temperature control range; from + 5° to 70°C (+/- 0.1°C)
- Variable rotation speed control from 5 to 20 RPM.



#### **SPECIFICATIONS**

• Rotator Motor Speed 2 to 20 RPM

Approximate Oven
 Dimensions
 Width: 17.4" (44.5 cm)
 Depth: 17.75" (39.5 cm)

 Approximate Chamber Dimensions Height: 14.5" (36.8 cm) Width: 12.5" (31.8 cm) Depth: 12.0" (30.5 cm) • Operating Temperature Range

• Weight

• Power Input

 Hybridization Chamber Rotator Rack + 5° to 70°C (+/- 0.1°C)

75 lbs (34.0 kg)

110-120 volts, 220-240 volts

Holds up to 24 hybridization chambers during hybridization



# SCANNING AND VISUALIZATION

## Flexibility for the array workflow

One critically important step in the microarray analysis workflow is

the generation of high-quality images. PerkinElmer's ScanRI scanner portfolio includes easy to use and reliable benchtop scanners that meet these criteria and offer flexibility for array users.

Combined with their compact benchtop designs, low background noise and high sensitivity, our ScanRI portfolio provides attractive solutions that accommodate different workflow requirements.



ScanRl – Single slide reader, 3 µm resolution	ScanRl HD – Single slide reader, 1 µm resolution	ScanRI HDA – Autoloader, 1 µm resolution
Weight 15.5 kg	Weight 28 kg	Weight 33 kg
Dimensions 278 x 457 x 369 mm	Dimensions 316 x 549 x 432 mm	Dimensions 322 x 656 x 439 mm
Confocal scanner with integrated autofocus	Confocal scanner with motorized focus adjustment	Confocal scanner with motorized focus adjustment
Laser excitation wavelengths of 532 nm and 635 nm	Laser excitation wavelengths of 532 nm and 635 nm	Laser excitation wavelengths of 532 nm and 635 nm
Simultaneous 2-color scanning	Simultaneous 2-color scanning	Simultaneous 2-color scanning
Scanning resolution from 3 $\mu m$ to 40 $\mu m$	Scanning resolution from 1 $\mu m$ to 40 $\mu m$	Scanning resolution from 1µm to 40µm
Suitable for scanning of all standard DNA microarray slides (25-26 mm X 75-76 mm X 0.9-1.2 mm)	Suitable for scanning of all standard DNA microarray slides (25-26 mm X 75-76 mm X 0.9-1.2mm)	Suitable for scanning of all standard DNA microarray slides (25-26 mm X 75-76 mm X 0.9-1.2 mm)
Single slide reader	Single slide reader	Autoloader for 24 slides



# THE POWER OF BACS-ON-BEADS

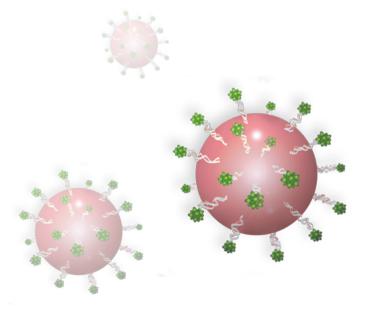
### BACs-on-Beads<sup>™</sup> (BoBs<sup>™</sup>)

is an exciting technology provided by PerkinElmer for rapid copy number detection

in selected genomic regions. By immobilizing BAC (bacterial artificial chromosome) derived DNA probes onto Luminex® xMAP® fluorescently coded beads, BACs-on-Beads technology supports high throughput molecular karyotyping in a microplate well, which in turn can lead to greater laboratory efficiency and better use of resources.

#### **BoBs technology**

BAC DNAs are large cloned sequences of human DNA with a typical length of 150-170 kb. When coupled to Luminex® xMAP® beads, the length of the BAC clones is a useful property as BAC derived DNA probes display a higher signal-to-noise ratio compared to smaller DNA probes such as oligonucleotides. This represents an important advantage specifically when working with samples such as amniotic fluid, CVS samples (Chorionic Villi samples), POC (Products of Conception) or DNA amplified from single cells typically used in PGS (Preimplantation Genetic Screening) applications.







# LUMINEX XIVAP

## Luminex® xMAP® Technology

is an established multiplexing technology utilizing approximately 5  $\mu m$  diameter polystyrene beads coded with

two different fluorescent dyes.

By using ten different concentrations of each of the two dyes it is possible to generate up to 100 bead types with distinct fluorescent signatures, that can be identified through excitation of the coded dyes by the Luminex 100/200™ instrument.

This allows for a high-throughput, cost-effective multiplexing technology to be implemented in cytogenetic laboratories.

### Streamlined data interpretation of the Copy Number Changes with the BoBsoft™ Analysis Software

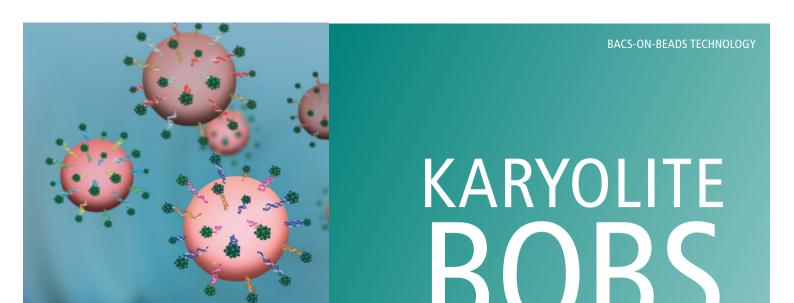
The BoBsoft™ analysis software utilizes the output files generated by the Luminex® 100/200™ instrument for data analysis and interpretation, and provides visualization of copy number changes in the genomic regions targeted by the BoBs products. The signal intensities from the sample and reference DNA are compared to provide a clear display of any copy number changes in the targeted regions.

#### **BOBS ASSAY CHARACTERISTICS**

- Robust assay with even low quality DNA derived from various sample types
- High-throughput analysis for costeffectiveness and reduced TAT
- Complete workflow in under 24 hours
- Easy and streamlined data analysis and interpretation with BoBsoft



Probe	Normali	zed Ratios	Threshold: 0,90-1,10 Threshold Y: 0,85-1,1	5
Probe	Sample/F	Sample/M	0.0 0.5 1.0 1.5	2
13C	1,01	0,99		
18C	1,00	0,99	4	
21C	0,96	0,98		
AUTO	1,01	1,01		
CDC	0,95	0,95		
DGS	1,03	1,04		
DIG	1,02	1,04		
LGS	0,67	0,66	1.1.1.1	
MDS	1,04	1,05		
PWS	1,00	0,99		
SMS	1,05	1,02		
WBS	0,97	0,99	<u> </u> -	
WHS	1,03	1,03		
xc	0,99	1,43		
YC	1,00	0,25		



### 24 Chromosomes in under 24 Hours

KaryoLite BoBs utilizes composite beads for aneuploidy analysis of all 24 chromosomes in under 24 hours

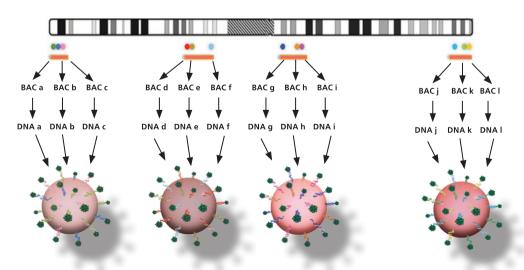
Based on the BACs-on-Beads technology, KaryoLite BoBs consists of BAC DNAs immobilized onto polystyrene microspheres. KaryoLite BoBs however utilizes a new concept of composite beads, whereby DNAs derived from three different BAC clones are combined on each bead type. This composite clone format expands the region of chromosomal DNA interrogated by each bead, and so allows detection of aneuploidies on all 24 chromosomes within one single experiment in as little as 24 hours.

Due to its high-throughput and cost-effectiveness, KaryoLite BoBs leans itself specifically to research applications such as the analysis of POC (Products of Conception) and PGS (Preimplantation Genetic Screening) samples.

#### KARYOLITE BOBS ASSAY CHARACTERISTICS

- Analysis of up to 92 samples within one single experiment
- . Cost-effectiveness due to high-throughput ability
- · Result reporting in under 24 hours
- · Easy data interpretation by use of BoBsoft

KaryoLite BoBs utilizes a new concept of composite beads combining three different BAC clones on each bead type.



KaryoLite BoBs reagents are for Research Use Only. Not for use in diagnostic procedures.



## SINGLE CELL WGA

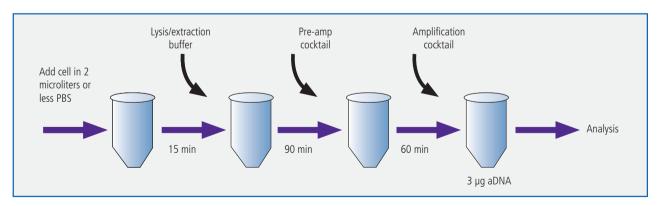
## PicoPLEX™ WGA Kit for whole genome amplification of single cells

Reliable DNA amplification is crucial for the accurate detection of

chromosomal aneuploidies when starting with low amounts of input DNA. This is specifically required in research applications such as PGS (Preimplantation Genetic Screening), where aneuploidy screening is typically performed on single cells or a small number of cells derived from blastomeres, blastocysts or polar bodies.

#### Rapid and robust one-tube protocol

The PicoPLEX<sup>TM</sup> WGA (Whole Genome Amplification) kit is designed and optimized specifically for single cell genomic DNA, and yields a reproducible representation of all sequences with low allele drop out. The amplification procedure is a simple 1-tube, 3-step, 3 hour protocol that produces 3-5 micrograms of amplified DNA sufficient for use of subsequent analyses such as KaryoLite BoBs or array CGH.



PicoPLEX™ WGA whole genome amplification process for single cell amplification

### **ORDERING INFORMATION**

Product Number	Product Name
chemagen products	
CMG-1072	chemagic DNA Blood Kit special, 50 μl, 96 samples
CMG-721	chemagic DNA Blood Kit special, 100 µl, 96 samples
CMG-746	chemagic DNA Blood Kit special, 250 μl, 96 samples
CMG-1091	chemagic DNA Blood Kit special, 400 μl, 96 samples
CMG-1086	chemagic DNA Blood Kit special, 1 ml, 24 samples
CMG-1097	chemagic DNA Blood Kit special, 2 ml, 24 samples
CMG-1074	chemagic DNA Blood Kit special, 4 ml, 24 samples
CMG-763-1	chemagic DNA Blood Kit special (4 ml elution tubes), 3 ml, 12 samples
CMG-763-2	chemagic DNA Blood Kit special (13 ml elution tubes), 3 ml, 12 samples
CMG-703-1	chemagic DNA Blood Kit special (13 ml elution tubes), 5 ml, 12 samples
CMG-703-2	chemagic DNA Blood Kit special (4 ml elution tubes), 5 ml, 12 samples
CMG-715	chemagic DNA Blood Kit special, 7 ml, 12 samples
CMG-704	chemagic DNA Blood Kit special, 10 ml, 12 samples
CMG-1030	chemagic DNA Blood Spot Kit special, 3 - 6 mm paperfilter punch outs, 96 samples
CMG-1081	chemagic DNA Saliva Kit special, 4 ml, 24 samples
CMG-1035	chemagic DNA Saliva Kit special, 12 samples
CMG-748	chemagic DNA Buccal Swab Kit special, 96 samples
CMG-756	chemagic DNA Cell12M Kit special, 12 samples
CMG-2002	Prepito DNA Blood250 Kit, 12 samples
CMG-2004	Prepito DNA Blood600 Kit, 6 samples
CMG-2027	Prepito FFPE Kit, 10 µm section, 12 samples
CMG-2010	Prepito DNA Tissue10 Kit, 10 mg tissue, 12 samples
CMG-108	chemagic SEQ Pure Kit, 10 - 20 µl
CMG-459	chemagic SEQ Pure Kit LH, 10 µl
CMG-458	chemagic SEQ Pure Kit LH, 20 μl
CMG-1096	chemagic Circulating NA Kit special, 1 ml, 12 samples
CMG-1090	chemagic Circulating NA Kit special, 4 ml, 24 samples
CMG-797	chemagic DNA Amniotic Fluid Kit special, 1 ml, 3 ml, 12 samples
CMG-1068	chemagic Epigenetic NA Extraction Kit special, 5 ml, 12 samples
CMG-1088	chemagic Epigenetic NA Extraction Kit special, 4 ml, 24 samples
CMG-1069	chemagic Epigenetic Bisulfite Purification Kit special, 500 μl, 96 samples
CMG-2034	Prepito DNA Cyto Pure Kit, 12 samples
CMG-2025	Prepito Circulating NA1k Kit, 6 samples
CMG-1083	chemagic RNA Blood Kit special, 12 samples
CMG-1084	chemagic RNA Blood Kit special, 24 samples
CMG-1093	chemagic RNA Saliva Kit special, 1 ml, 12 samples
CMG-1031	chemagic mRNA/gDNA Kit special, 2 ml blood or bone marrow, 12 samples
CMG-360	chemagic 360 instrument
2022-0020	chemagic Prepito-D instrument

Product Number	Product Name
PicoPLEX™WGA	
4504-0010	PicoPLEX WGA Kit
BACs-on-Beads	
1014-0020	Luminex® 200 w/ xPONENT®
4500-0020	Constitutional BoBs
4501-0010	KaryoLite™ BoBs™
CGX™	
1013-0010	ScanRI Scanner
1013-0040	ScanRI HD Scanner
1013-0030	ScanRI HDA Scanner
1013-0020	ScanRI PC for ScanRI
1013-0050	ScanRI PC for ScanRI HD and ScanRI HDA
5016-0010	Genoglyphix SW
4113-0010	CGX™ HD/SNP Gasket Slides-100
4114-0010	CGX™ Gasket Slides-100
4115-0010	CGX™ HD/SNP Gasket Slides-20
4116-0010	CGX™ Gasket Slides-20
4117-0010	CGX™ Ozone-barrier slide covers
4118-0020	CGX™ (2 slides per pack)
4119-0020	CGX™ (4 slides per pack)
4122-0020	CGX™ HD (2 slides per pack)
4123-0020	CGX™ HD (4 slides per pack)
4124-0020	CGX™ SNP (2 slides per pack)
4125-0020	CGX™ SNP (4 slides per pack)
1015-0020	CGX <sup>™</sup> Hybridization chamber, stainless
1015-0040	CGX™ Hybridization oven
1015-0030	CGX™ Hybridization oven rotator
4130-0010	CGX™ Cot-1 Human DNA
4131-0010	CGX <sup>™</sup> Oligo aCGH Hybridization Kit
4132-0010	CGX™ Oligo aCGH Wash Kit
4135-0010-P4	CGX <sup>™</sup> DNA Labeling Kit – Purification Columns
4134-0010	CGX <sup>™</sup> Stabilization and Drying Solution
4135-0010	CGX™ DNA Labeling Kit

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