



SurePrint Community Design Prenatal EasyChip 8x15K

Simplify your prenatal research
with a targeted approach

Advantages of the design

- Clinically relevant targets
- Curated low resolution backbone
- Higher coverage on subtelomeric regions
- Minimal starting DNA requirements
- Designed to limit VOUS and IF for easier interpretation

Chromosomal microarray for prenatal research

The demand for prenatal in-depth analyses over standard karyotype testing has dramatically increased in recent years. Chromosome Microarray Analysis (CMA) provides information on small rearrangements that are often overlooked by banded metaphases.

CMA methods to detect genomic imbalances (e.g., copy number variations (CNVs)) are widely used in prenatal research after an abnormal ultrasound result, as this technology provides a deeper characterization of chromosomal anomalies. Microarrays have recently been suggested as Tier I tests after noninvasive screening by the American Society of Human Genetics¹, illustrating the advantage of CMA over next generation sequencing (NGS) for this application.

SurePrint CD Prenatal 8x15K

SurePrint Community Design (CD) Prenatal EasyChip 8x15K is an oligonucleotide array design that allows highly focused analyses of any prenatal samples while minimizing incidental findings (IF) and variants of unknown significance (VOUS).

This new microarray enables both high-resolution investigation of 43 syndromic regions (Table1) and low-resolution screening of the whole genome including subtelomeric regions.

The 43 genomic regions critical for prenatal analysis have been selected to map highly penetrant (>70%) morbid conditions². The arrays are easily analyzed since they target well-characterized genes/regions and present deletions/duplications as main etiological mechanisms^{3,4}.

Although not all subtelomeric regions have been associated with distinct conditions, they have all been included in the microarray because of their high recombination rate, disposition towards imbalances, and frequent association with intellectual disability⁵⁻⁷. SurePrint CD Prenatal EasyChip 8x15K also provides a 3 Mb functional resolution at the genomic backbone level in order to allow integrated karyotype analyses. This streamlines the cytogenetic workflow, especially when the quality of metaphases is poor.

Table 1. List of syndromic regions selected to be covered by the design.

Syndromic Regions	Incidence	Critical Genes
1p36 deletion syndrome	1/5,000	/
1q41q42 microdeletion syndrome	unknown	<i>DISP1</i>
2p15-16.1 microdeletion syndrome	unknown	<i>BCL11A</i>
2q23.1 microdeletion syndrome	unknown	<i>MBD5, EPC2</i>
2q33.1 deletion (Glass syndrome)	unknown	<i>STAB2</i>
2q37 deletion syndrome	<1/10,000	<i>HDAC4</i>
3pter-p25 deletion syndrome	unknown	<i>CNTN4, ITPR1, SRGAP3, VHL</i>
3q29 deletion/duplication syndrome	unknown	<i>FBXO45, PAK2, DLG1</i>
4p16.3 deletion syndrome (Wolf-Hirschhorn)	1/20,000 - 1/50,000	<i>LETM1, WHSC1</i>
4q21 deletion syndrome	unknown	<i>PRKG2, RASGEF1B</i>
5p deletion syndrome (Cri du chat)	1/20,000 - 1/50,000	<i>CTNND2, TERT</i>
5q14.3 deletion syndrome	unknown	<i>MEF2C</i>
5q35 deletion syndrome (Sotos)	1-9/100,000	<i>NSD1</i>
6q13-q14 deletion syndrome	unknown	<i>COL12A1</i>
7q11.23 deletion syndrome (Williams-Beuren)	1/10,000	<i>ELN</i>
8p23.1 deletion syndrome	unknown	<i>GATA4</i>
8q21.11 Microdeletion Syndrome	unknown	<i>ZFXH4, PEX2</i>
8q24.1 deletion syndrome (Langer-Giedion)	unknown	<i>TRPS1, EXT1</i>
9q34.3 deletion syndrome (Kleefstra)	unknown	<i>EHMT1</i>
10p14p13 deletion syndrome (DiGeorge type 2)	unknown	<i>GATA3</i>
11p13 deletion syndrome (WAGR)	unknown	<i>PAX6, WT1</i>
11p11.2 deletion syndrome (Potocki-Shaffer)	unknown	<i>ALX4</i>
11q deletion syndrome (Jacobsen)	1/100,000	/
14q12 microdeletion syndrome	unknown	<i>FOXP1</i>

Syndromic Regions	Incidence	Critical Genes
15q11q13 deletion syndrome (Prader-Willi)	1/25,000	<i>SNRPN</i>
15q11q13 deletion syndrome (Angelman)	1/10,000 - 1/20,000	<i>UBE3A</i>
15q24 deletion/duplication syndrome	unknown	/
16p deletion syndrome (ATR-16)	unknown	<i>HBA1, HBA2</i>
16q24.1 microdeletion syndrome	unknown	<i>FOXF1, FOXC2</i>
17p13.3 deletion syndrome (Miller dieker)	unknown	<i>PAFAH1B1, YWHAE</i>
17p11.2 deletion syndrome (Smith-Magenis)	1/25,000	<i>RAI1</i>
17p11.2 duplication syndrome (Potocki Lupski)	unknown	<i>RAI1</i>
17q11.2 deletion/duplication syndrome	unknown	<i>NF1, SUZ12</i>
17q21.31 deletion syndrome (Koolen-De Vries)	1/16,000	<i>KANSL1</i>
17q23.1-q23.2 deletion syndrome	unknown	<i>TBX2, TBX4</i>
19q13.11 deletion syndrome	unknown	<i>LSM14A, UBA2</i>
Down Syndrome critical region (21q22.12q22.2)	1/650 - 1,000	/
22 partial tetrasomy (Cat eye)	1/50,000 - 1/150,000	/
22q11.2 deletion syndrome (DiGeorge)	1/2,000 - 1/4,000	<i>HIRA, TBX1</i>
22q11.2 distal deletion syndrome	unknown	<i>MAPK1</i>
Xp11.3 deletion syndrome	unknown	<i>RP2</i>
Xp11.22 microduplication syndrome	unknown	<i>HUWE1</i>
Xq12 deletion/duplication (OPHN1)	unknown	<i>OPHN1</i>
Xq22.3 deletion syndrome (AMME COMPLEX)	unknown	<i>COL4A5, ACS4</i>
Xq28 duplication syndrome	unknown	<i>MECP2</i>

Design details

SurePrint CD Prenatal EasyChip 8x15K is composed of 3 main probe groups that target different genome regions at defined resolutions.

Group 1 – Target Regions: 43 regions identified to be critical for prenatal analysis, targeted by high quality probes with average median probe spacing of 22.7 Kb. Using a minimum of 7 probes to call an aberration, this design provides a functional resolution of 150 Kb.

Group 2 – Subtelomeric: probes have been selected within the SureDesign CGH HD database using an average probe spacing of 50 Kb. Final median probe spacing is 44.9 Kb, enabling a functional resolution of 300 Kb for all subtelomeric regions.

Group 3 – Backbone: in order to obtain an average functional resolution of 3 Mb along the entire genome, all probes were selected using an average probe spacing of 500 Kb and similarity score filter on. The median probe spacing was 453.5 Kb with a total coverage >99.8% and >99.5% of high-quality probes.

Developed by an expert

This array has been developed by Professor Antonio Novelli, an expert cytogeneticist with years of experience in cytogenetics and in CGH applied to pre- and post-natal clinical research.

Professor Novelli is currently the Director of the Medical Genetic Unit at the Bambino Gesù Pediatric Hospital in Rome, Italy.

Order information

Product Description	Part Number	# Samples/Kit
SurePrint Community Design Prenatal EasyChip 8x15K	G5988A	8
SureTag Complete DNA Labelling kit	5190-4240	50
SureTag Purification Columns kit	5190-3391	50
Co-1 Human DNA	5190-3393	312
Hybridization Gasket Slide kit - 8 microarrays per slide format; 5 gasket slides/pack	G2534-60014	40
Oligo aCGH/ChIP-on-chip Hybridization kit	5188-5200	200
Oligo aCGH/ChIP-on-chip Wash Buffer kit	5188-5226	320

Go to <http://www.agilent.com/genomics> to see all available kit configurations

Note: Agilent has not performed verification and validation on these arrays.

Citations

1. Monaghan, K.G.; Leach N.T.; Pekarek, D.; Prasad, P.; Rose, N.C. The use of fetal exome sequencing in prenatal diagnosis: a points to consider document of the American College of Medical Genetics and Genomics (ACMG). *Genetics in Medicine*. **2020**, *22*: 675–680.
2. Alesi, V.; Bernardini, L.; Goidin, D.; Canestrelli, M.; Dentici, M.L.; Barrano, G.; Giuffrida, M.G.; Nardone, A.M.; Postorivo, D.; Laino, L.; Genesio, R.; Dallapiccola, B.; Novelli, A. Easychip 8x15k: A New Tool for Detecting Chromosome Anomalies in Low Risk Pregnancies, Supporting and Integrating Standard Karyotype. *J. Genet. Syndr. Gene Ther.* **2015**, *7*: 277.
3. Girirajan, S.; Rosenfeld, J.A.; Coe, B.P.; Parikh, S.; Friedman, N.; Goldstein, A.; Filipink, R.A.; McConnell, J.S.; Angle, B.; Meschino, W.S.; Nezarati, M.N.; Asamoah, A.; Jackson, K.E.; Gowans, G.C.; Martin, J.A.; Carmany, E.P.; Stockton, D.W.; Schnur, R.E.; Penney, L.S.; Martin, D.M.; Raskin, S.; Leppig, K.; Thiese, H.; Smith, R.; Aberg, E.; Niyazov, D.M.; Escobar, L.F.; El-Khechen, D.; Johnson, K.D.; Lebel, R.R.; Siefkas, K.; Ball, S.; Shur, N.; McGuire, M.; Brasington, C.K.; Spence, J.E.; Martin, L.S.; Clericuzio, C.; Ballif, B.C.; Shaffer, L.G.; Eichler, E.E. Phenotypic heterogeneity of genomic disorders and rare copy-number variants. *N. Engl. J. Med.* **2012**, *367*: 1321-1331.
4. Rosenfeld, J.A.; Coe, B.P.; Eichler, E.E.; Cuckle, H.; Shaffer, L.G. Estimates of penetrance for recurrent pathogenic copy-number variations. *Genet. Med.* **2013**, *15*: 478-481.
5. De Vries, B.B.; Winter, R.; Schinzel, A.; van Ravenswaaij-Arts, C. Telomeres: a diagnosis at the end of the chromosomes. *J. Med. Genet.* **2003**, *40*: 385-398.
6. Ravnan, J.B.; Tepperberg, J.H.; Papenhausen, P.; Lamb, A.N.; Hedrick, J.; Eash, D.; Ledbetter, D.H.; Martin, C.L. Subtelomere FISH analysis of 11 688 cases: An evaluation of the frequency and pattern of subtelomere rearrangements in individuals with developmental disabilities. *J. Med. Genet.* **2006**, *43*: 478-489.
7. Ballif, B.C.; Sulpizio, S.G.; Lloyd, R.M.; Minier, S.L.; Theisen, A.; Bejjani, B.A.; Shaffer, L.G. The clinical utility of enhanced subtelomeric coverage in array CGH. *Am. J. Med. Genet.* **2007**, *143*: 1850-1857.

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Agilent has not performed verification and validation on this design.

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