



KaryoNIM® Prenatal

KaryoNIM Prenatal® is an aCGH-based platform, designed by our company that can be used to detect simultaneously, the presence or absence (amplifications/deletions) of genetic and chromosomal alterations, responsible of the 96 most common genetic syndromes, with a resolution 10 times higher than the traditional karyotype.

Why to use KaryoNIM® for prenatal diagnosis?

High resolution and design oriented to the genetic diagnosis

KaryoNIM® uses aCGH technology, and includes 15000 oligonucleotide probes along the whole human genome, with 1 probe every 300kb. This gives a median detection resolution of 1 - 1.5 Mb, which is 10 times higher than conventional karyotype.

KaryoNIM® is focused in the detection of genetic alterations related to genetic syndromes. In the critical regions, the resolution is 50 times higher than the conventional karyotype. With this design, we avoid unnecessary information in sensitive samples, like prenatal samples, focusing the analysis on the regions associated with known pathologies.

Easy and Reproducible Protocol

KaryoNIM® does not need cell culture to obtain metaphase cells. It only needs DNA from the sample. The minimum amount of DNA is 100ng, being 200ng the optimal. DNA quality is crucial, so special care must be taken in handling the sample, specially the DNA extraction step.

The DNA of the sample will be compared with a commercial DNA control. Both samples will be fluorescently labeled in different colors and hybridized on to the KaryoNIM® platform, then scanned and the data acquired and analyzed.

Reliable and Fast Results

It takes 5 to 10 days since we get the sample till the technical report is ready. The analysis is performed by the bioinformatics team, and is based on standards of quality and quantitative statistic parameters. The detection of the alterations on the sample is automatized, so it doesn't depend on the staff experience.

KaryoNIM® let us see the alterations present, or not in the patient DNA, at the level of duplications, deletions or amplifications. This analysis allows us to detect alterations in mosaic of up to 30% in the sample.



REPORT

The report includes a clear answer (positive or negative) for all of the syndromes included specifically in the array. Additionally, any other chromosomal alteration that implies genomic gain or loss higher than 1.5 Mb, and that is present in a minimum mosaic of 30% of the total sample, will be present in the report.

PRACTICAL INFORMATION

KaryoNIM® is already available for our clients upon request.

In all cases KaryoNIM® is a technology that complements conventional karyotype. It can substitute prenatal FISH by all means, and even expands the information, detecting simultaneously the 96 more common syndromes that involve microdeletion/duplication. NIMGenetics eventually can take care of performing the karyotype of the sample.

For additional information, procedures and prices, please contact NIMGenetics.

KaryoNIM Prenatal® es una plataforma basada en aCGH diseñada por nuestra empresa, que se puede utilizar para detectar simultáneamente, la presencia o ausencia (amplificaciones o deleciones) de alteraciones genéticas y cromosómicas, responsables de los 96 síndromes genéticos más comunes, con una resolución 10 veces mayor que el cariotipo convencional.

LIST OF SYNDROMES INCLUDED IN karyoNIM® prenatal

Chromosome	List of Syndromes included in KaryoNIM®	# OMIM	Cytoband	Detected	Healthy
1	Monosomy 1p36 Syndrome	#607872	1pterp36		✓
1	Chromosome 1q43q44 Microdeletion Syndrome	#612337	1q43q44		✓
2	Feingold Syndrome	#164280	2p24		✓
2	Hypotonia-cystinuria Syndrome	#606407	2p16.3		✓
2	Holoprosencephaly 2	#157170	2p21		✓
2	Microdeletion 2p16.1p15 Syndrome	#612513	2p16.1p15		✓
2	Joubert Syndrome 4 / Nephronophthisis 1	#609583	2q13		✓
2	Split-hand/foot Malformation 5	606708%	2q31.1		✓
2	Sinplidactilia 1	186000%	2q31.1		✓
2	Microdeletion 2q31 Syndrome	#612345	2q31		✓
2	Microdeletion 2q32q33 Syndrome	#612313	2q32-33		✓
2	Holoprosencephaly 6	605934%	2q37.1-q37.3		✓
2	Braquidactyly – Mental Retardation Syndrome	600430%	2q37.3qter		✓
3	Dandy-Walker Syndrome	220200%	3q24		✓
3	Microphthalmia Syndromic 3	#206900	3q26		✓
3	Split-hand/foot Malformation 4	#605289	3q28		✓
3	Microdeletion 3q29 Syndrome	609425%	3q29qter		✓
4	Wolf-Hirschhorn Syndrome	#194190	4p16.3		✓
4	Axenfeld Rieger Syndrome	#180500	4q25		✓
5	Cri-du-chat Syndrome	#123450	5pterp15.33		✓
5	Cri-du-chat Syndrome, Distal Región	#123450	5p15.2		✓
5	Cornelia de Lange Syndrome	#122470	5p13.2		✓
5	Sotos Syndrome	#117550	5q35.2		✓
6	Microdeletion 6pterp24 Syndrome	#612582	6pter6p24		✓
6	Cleidocraneal Dysplasia	#119600	6p21.1		✓
6	Prader-Willi-Like Syndrome	#176270	6q16.3		✓
7	Saethre-Chotzen Syndrome	#101400	7p21		✓
7	Creig Cephalopolysyndactyly Syndrome	#175700	7p13		✓
7	Williams Beuren Syndrome	#194050	7q11.23		✓
7	Williams Beuren Duplication Syndrome	#609757	7q11.23		✓
7	Split-Hand/foot Malformation 1	183600%	7q21.3		✓
7	Holoprosencephaly 3	#142945	7q36.3		✓
8	Charge Syndrome	#214800	8q12.1		✓
8	Langer Giedon Syndrome	#150230	8q23q24		✓
8	Triconofaringeo 1 Syndrome	#190350	8q23q24		✓
9	46,XY Gonadal Dysgenesis Delección, Complete or Partial with 9q24.3 Deletion	#154230	9q24.3		✓
9	Microdeletion 9p Syndrome	#158170	9p		✓
9	Holoprosencephaly 7	#610828	9q22.3		✓
9	Naill-Patella Syndrome	#161200	9q34.1		✓
9	Microdeletion 9q34.3	#610253	9q34.3		✓
10	Hipoparatiroidismo, sordera sensorineural y enfermedad renal	#146255	10p15		✓
10	Digeorge 2 Syndrome	601362%	10p14		✓
10	Digeorge 2 Syndrome, Nebulette Region	601362%	10p13		✓
10	Microdeletion 10q23 Syndrome	612242	10q23		✓
10	Split-Hand/Foot Maldormation 3	600095%	10q24.32		✓
11	Beckwith-Wiedemann Syndrome	#130650	11pter		✓
11	Microdeletion 11p15p14 Syndrome	606528	11p15p14		✓
11	WAGR Syndrome (Including Wilms Tumor)	#194072	11p13		✓
11	Potocki-Shaffer Syndrome	#601224	11p11.2		✓
11	Jacobsen Syndrome	#147791	11q25qter		✓
12	Pallister-Killian Syndrome	#601803	12pterpcen		✓
13	Patau Syndrome (Chromosome 13 Trisomy)	-	tri13		✓
13	Holoprosencephaly 5	#609637	13q32.3		✓
14	Microphthalmia Sindromic 6; MCOPS6	#607932	14q22q23		✓

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15	Prader-Willi Syndrome/ Angelman Syndrome	#176270	15q12		✓
15	Diaphragmatic Hernia diafragmática, Congenital	142340%	15q26qter		✓
16	Telomere 16pter-p13.3 Alpha thalasemia/Mental Retardation Syndrome	#141750	16pter-p13.3		✓
16	16p13.3" Polycystic kidney disease, Infantile Severe with Tuberos Sclerosis	#600273	16p13.3		✓
16	Rubinstein-Taybi Syndrome	#180849	16p13.3		✓
16	Microdeletion 16p13.3Syndrome (RTSS)	#610543	16p13.3		✓
17	Miller-Dieker Lissencephaly Syndrome	#247200	17p13.3		✓
17	17p13.3 Chromosome Duplication Syndrome	#613215	17p13.3		✓
17	Charcot-Marie Tooth Syndrome 1A	#118220	17p11.2		✓
17	Neuropathy, Hereditary, UIT Liability to pressure palsies, HNPP	#162500	17p11.2		✓
17	Smith-Magenins Syndrome	#182290	17p11.2		✓
17	Potocki-Lupski Syndrome	#610883	17p11.2		✓
17	NF1 Microdeletion Syndrome	#162200	17q11.2		✓
17	Microdeletion 17q21.31 Syndrome	#610443	17q21.31		✓
17	Campomelic Dysplasia	#114290	17q24.3		✓
18	Edwards Syndrome	-	Tri18		✓
18	Holoprosencephaly 4	#142946	18p11.31		✓
18	Pitt-Hopkins Syndrome	#610954	18q21.2		✓
18	Microdeletion 18q Syndrome	#601808	18qter		✓
18	Aural Atresia, Congenital	#607842	18qter		✓
19	Microdeletion 19q13.1 Syndrome	#613026	19q13.1		✓
20	Alagille Syndrome 1	#118450	20p12.2		✓
21	Down Syndrome	#190685	Tri21		✓
21	Critic Region Down Syndrome	#190685	21q22		✓
22	Holoprosencephaly 1	236100%	21q22.3		✓
22	Cat-Eye Syndrome	#115470	22q11.1		✓
22	Digeorge Syndrome / Velocardiofacial Syndrome	#188400	22pterq11.2		✓
22	Microdeletion 22q11.2 Syndrome, Distal	#611867	22q11.2		✓
22	Microdeletionn 22q13.3 Syndrome	#606232	22q13.3qter		✓
X	Turner Syndrome	-	DelX		✓
X	Triple X Syndrome	-	TriX		✓
X	Klinefelter (XXY) Syndrome	-	XXY		✓
X	Complicated X-linked Ictiosis Due to Contiguous Gene Deletion Syndrome	#308100	Xp22.31		✓
X	Duchenne Muscular Distrophy	#310200	Xp21.2		✓
X	Microdeletion Xp11.3 Syndrome	#300578	Xp11.3		✓
X	Xp11.23p11.22 Duplication Syndrome	#300801	Xp11.23p11-22		✓
X	Mental Retardation, X-linked, Syndromic, Turner Type	#300706	Xp11.2		✓
X	Mental Retardation, X-linked with panhypopituitarism	#300123	Xq26.3		✓
X	Contiguous ABCD1/DXS1375E Microdeletion Syndrome	#300475	Xq28		✓
X	MECP2 Duplication Syndrome	#300260	Xq28		✓
Y	SRY Mapping	-	Yq11.3		✓
Y	XXX Syndrome	-	XXY		✓