



BIOARRAY
Diagnóstico Genético



www.bioarray.es | +34 966 682 500

1. CGH MICROARRAY	04
1.1. Cytoarray	05
1.2. Cytoarray Plus	06
1.3. Prenatal Diagnosis Cytoarray	07
1.4. Cytoarray UPD	52
1.5. DACrA	53
1.6. High Density CGH Microarray. 400K y 1M	55



B I O A R R A Y
Diagnóstico Genético



BA

BIOARRAY
Diagnóstico Genético

1

CGH
MICROARRAY

1. CGH MICROARRAY

1.1. CYTOARRAY

It is a high-resolution comparative genomic hybridization microarray designed specifically for clinical genetics and cytogenetics.

It detects DNA copy number variations in patients with idiopathic mental retardation, autism and other congenital abnormalities. It enables to determine whether the patient has a disease or syndrome associated to the presence of microdeletions or microduplications in defined DNA regions (see syndrome list attached).

Based on the design made by the ISCA (The International Standards for Cytogenomic Arrays Consortium), international reference for cytogenetics laboratories, it has become a tool to identify copy number variations alongside the whole genome.

Cytoarray, with more than sixty thousand probes, has been specially developed to support numerous cytogenetic syndromes by accurate detection of microdeletions and microduplications, improving outcomes against conventional cytogenetic techniques.

CYTOARRAY CAN DETECT

- Aneuploidies.
- Alterations sized about 48Kb in nearly 500 target areas.
- Over 400 syndromes (one probe every 31Kb)
- 233 genes associated with autism, mental retardation, congenital heart and eye diseases.
- Subtelomeric bands (one probe every 75 Kb)

To summarize, it is a powerful diagnostic tool that provides more information than traditional methods in order to acquire a more effective diagnosis.

1.2. CYTOARRAY PLUS

With a similar design to Cytoarray, but an increased definition and resolution, Cytoarray Plus contains a hundred and eighty thousand probes. It has been specially developed to identify very small changes in multiple genes related to pathology.

TECHNICAL CHARACTERISTICS

- Probe Density: 1 probe every 25Kb. Probe resolution: 1 probe every 14kb, contributing an average of 6 probes per gene.
- 501 regions of genes associated with autism, mental retardation, congenital heart and eye diseases.



1.3. PRENATAL DIAGNOSIS CYTOARRAY

It is a microarray developed specifically for prenatal diagnosis that can detect nearly 500 syndromes associated with 233 genes. Because of its whole genome coverage, it is a perfect tool for prenatal diagnosis. Besides, results are reported within a week.

APPLICATIONS

- Karyotypes with abnormalities.
- Abnormal echography.
- Fetal death or recurrent loss of gestation.
- History of children with genetic conditions.
- Maternal anxiety.



SYNDROME TABLE

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREGNATAL
1p36.33	Telomere			•	•	•
1p36.33	SKI	1p36 Microdeletion	607872	•	•	•
1p36.32	TP73	1p36 Microdeletion	607872	•	•	•
1p36.13	CLCNKA	Bartter 4B	613090	•	•	•
1p36.13	CLCNKB	Bartter 3	607364	•	•	•
1p32	BSND	Bartter 4A	602522		•	
1p32.3	BSND			•	•	•
1p31.3	NFIA			•	•	•
1p31.3	DIRAS3			•	•	•
1p21.1	COL11A1	Stickler syndrome, type II; STL2	604841	•	•	•
1p12	Centromere			•	•	•
1q21.1	Centromere			•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PRENATAL
1q21.2	GJA5	Atrial fibrillation		•	•	•
1q21.2	GJA8	Cataract-microcornea syndrome	116150	•	•	•
1q25.2	LHX4	Pituitary hormone deficiency, combined 4; CPHD4	262700	•	•	•
1q32.2	IRF6	Van der Woude syndrome 1; VWS1	119300	•	•	•
1q41	DISP1	Fryns 1q41	229850	•	•	•
1q42.2	DISC1	Autism	209850	•	•	•
1q42.3	TBCE	Hypoparathyroidism-retardation-dysmorphism syndrome, HRD	241410	•	•	•
1q44	Telomere			•	•	•
2p25.3	Telomere			•	•	•
2p24.3	MYCN	Feingold	164280	•	•	•
2p23.1	SRD5A2			•	•	•
2p22.1	SOS1			•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDNATAL
2p21	SLC3A1	Cystinuria with mitochondrial disease	606407	•	•	•
2p21	PREPL	Cystinuria with mitochondrial disease	606407	•	•	•
2p21	SIX3	Holopresencaphaly 2	157170	•	•	•
2p16.3	NRXN1	Autism	209850	•	•	•
2p15	MicroDeletionRegion	2p15-p16.1 Microdeletion		•	•	•
2p15	Microdel. Region			•	•	•
2p11.2	Centromere			•	•	•
2q11.1	Centromere			•	•	•
2q13	NPHP1			•	•	•
2q14.2	GLI2	Holoprosencephaly 9	610829	•	•	•
2q21.1	CFC1	Heterotaxy, visceral, 2, autosomal; HTX2	605376	•	•	•
2q21.1	CFC1	Heterotaxy, visceral, 3, autosomal; HTX3	605377	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDNATAL
2q22.3	ZEB2	Mowat-Wilson syndrome	235730	•	•	•
2q24.2	SLC4A10	Autism	209850	•	•	•
2q24.3	SCN1A	Generalized epilepsy with febrile seizures plus, type 2; GEFSP2	604403	•	•	•
2q31.1	SLC25A12	Hypomyelination, global, cerebral	612949	•	•	•
2q31.1	DLX1			•	•	•
2q31.1	DLX2			•	•	•
2q31.1	EVX2			•	•	•
2q31.1	HOXD genes			•	•	•
2q31.1	HOXD13			•	•	•
2q31.1	HOXD9			•	•	•
2q33.1	SATB2	Cleft palate	119540	•	•	•
2q33.1	BMPR2	Pulmonary hypertension, primary 1; PPH1,	178600	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PRENATAL
2q36.1	PAX3	Waardenburg syndrome, type 1; WS1	193500	•	•	•
2q37.3	HDAC4	2q37.3 Monosomy		•	•	•
2q37.3	Telomere			•	•	•
10p15.3	Telomere			•	•	•
3p26.3	Telomere			•	•	•
3p26.3	CNTN4	Autism	209850	•	•	•
3p25.3	VHL	Von Hippel-Lindau syndrome; VHL	193300	•	•	•
3p25.2	RAF1	Noonan syndrome 5; NS5	611553	•	•	•
3p24.1	TGFBR2	Loeys-Dietz	610380	•	•	•
3p21.31	TDGF1	Teratocarcinoma-derived growth factor; TDGF1	187395	•	•	•
3p14.1	MITF	Waardenburg syndrome, type 2A; WS2A	193510	•	•	•
3p11.1	Centromere			•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDNATAL
3q11.1	Centromere			•	•	•
3q21.1	CASR	Bartter with autosomal dominant hypocalcemia	601199	•	•	•
3q22.3	FOXL2	Blepharophimosis	110100	•	•	•
3q24	ZIC4	Dandy-Walker	220200	•	•	•
3q24	ZIC1	Dandy-Walker	220200	•	•	•
3q26.33	SOX2	Microphthalmia, syndromic 3; MCOPS3	206900	•	•	•
3q28	TP73L	Split-hand/foot malformation 4; SHFM4	605289	•	•	•
3q29	PAK2	3q29 Microdeletion	609425	•	•	•
3q29	Telomere			•	•	•
4p16.3	Telomere			•	•	•
4p16.3	LETM1			•	•	•
4p16.3	WHSC1			•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDICTIVE
4p16.3	WHSC2			•	•	•
4p16.2	MSX1	Cleft lip	608874	•	•	•
4p11	Centromere			•	•	•
4q12	Centromere			•	•	•
4q22.1	PKD2	Polycystic kidney disease 2; PKD2	613095	•	•	•
4q25	PITX2			•	•	•
4q35.2	Telomere			•	•	•
5p15.33	Telomere			•	•	•
5p15.33	TERT	Cri-du-Chat	123450	•	•	•
5p13.2	NIPBL	Cornelia de Lange	122470	•	•	•
5p12	Centromere			•	•	•
5q11.1	Centromere			•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDNATAL
5q22.2	APC	Gardner	175100	•	•	•
5q23.2	LMNB1	Adult-onset autosomal dominant leukodystrophy	169500	•	•	•
5q32	SPINK1	Chronic pancreatitis	167800	•	•	•
5q32	TCOF1	Deafness	154500	•	•	•
5q35.1	FBXW11	Holoprosencephaly and preaxial polydactyly	264480	•	•	•
5q35.1	NKX2-5	Atrial septal defect with atrioventricular conduction defects	108900	•	•	•
5q35.2	MSX2	Craniosynostosis	604757	•	•	•
5q35.2	NSD1			•	•	•
5q35.3	Telomere			•	•	•
6p25.3	Telomere			•	•	•
6p25.3	FKHL7(FOXC1)	6p24 Deletion	612852	•	•	•
6p21.33	CYP21A2	Congenital adrenal hyperplasia (CAH)	201910	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDAT
6p21.32	SYNGAP1	Mental retardation, autosomal dominant 5; MRD5	612621	•	•	
6p21.1	VEGFA	Microvascular complications of diabetes, susceptibility to, 1; MVCD1	603933	•	•	•
6p21.1	RUNX2	Cleidocranial dysplasia	119600	•	•	•
6p12.1	Centromere			•	•	•
6q11.1	Centromere			•	•	•
6q16.3	SIM1	Obesity	601665	•	•	•
6q16.3	GRIK2	Mental retardation, autosomal recessive 6; MRT6	611092	•	•	•
6q23.3	AHI1	Joubert 3	608692	•	•	•
6q24.2	ZAC(PLAGL1)	Diabetes mellitus, transient neonatal, 1	601410	•	•	•
6q24.2	HYMAI	Diabetes mellitus, transient neonatal, 1	601410	•	•	•
6q25.3	IGF2R_ICregion			•	•	•
6q27	Telomere			•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREGNATAL
7p22.3	Telomere			•	•	•
7p21.1	TWIST1			•	•	•
7p14.1	GLI3	Greig cephalopolysyndactyly	175700	•	•	•
7p12.1	GRB10			•	•	•
7p11.2	Centromere			•	•	•
7q11.21	Centromere			•	•	•
7q11.23	ELN			•	•	•
7q11.23	LIMK1			•	•	•
7q11.23	GTF2IRD1				•	
7q11.23	GTF2I				•	
7q11.23	NCF1	Granulomatous disease, chronic, cytochrome b-positive	233700	•	•	•
7q21.3	COL1A2	Ehlers-danlos	225320	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDICTIVE
7q21.3	SGCE	Dystonia-11	159900	•	•	•
7q21.3	PEG10			•	•	•
7q21.3	PEG10_ICregion	Dystonia-11	159900	•	•	•
7q21.3	PPP1R9A			•	•	•
7q21.3	SHFM1			•	•	•
7q21.3	DLX5	Split-hand/foot malformation 1 with sensorineural hearing loss; SHFM1D	220600	•	•	•
7q22.1	NPTX2			•	•	•
7q22.1	RELN	Lissencephaly 2; LIS2	257320	•	•	•
7q31.1	FOXP2	Speech-Language disorder 1; SPCH1	602081	•	•	•
7q31.2	MET	Autism	209850	•	•	•
7q31.32	CADPS2	Autism	209850	•	•	•
7q32.2	CPA4			•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDNATAL
7q32.2	MEST			•	•	•
7q32.3	KLF14			•	•	•
7q34	PRSS1	Mental retardation		•	•	•
7q35	CNTNAP2	Autism	209850	•	•	•
7q36.3	EN2	Autism	209850	•	•	•
7q36.3	SHH	Holoprosencephaly 3	142945	•	•	•
7q36.3	LMBR1	Polydactyly, preaxial II; PPD2	174500	•	•	•
7q36.3	HLXB9(MNX1)	Currarino	176450	•	•	•
7q36.3	Telomere			•	•	•
8p23.3	Telomere			•	•	•
8p23.3	DLGAP2			•	•	•
8p23.1	MFHAS1	diGeorge 1	188400	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDATAL
8p23.1	GATA4	diGeorge 1	188400	•	•	•
8p23.1	CTSB			•	•	•
8p23	CTSB	Kabuki	147920	•	•	•
8p11.22	FGFR1	Kallmann 2	147950	•	•	•
8p11.1	Centromere			•	•	•
8q11.1	Centromere			•	•	•
8q12.1	CHD7	Charge	214800	•	•	•
8q13.3	EYA1	Branchio-oto-renal	113650	•	•	•
8q21.13	IMPA1			•	•	•
8q23.1	ZFPM2(FOG2)	Congenital heart	610187	•	•	•
8q23.3	TRPS1			•	•	•
8q24.11	EXT1	Langer-Giedion	150230	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREGNATAL
8q24.3	KCNK9	Birk-Barel mental retardation dysmorphism syndrome	612292	•	•	•
8q24.3	Telomere			•	•	•
9p24.3	Telomere			•	•	•
9p24	DMental RetardationT1	9p Deletion	158170	•	•	•
9p24.3	DMRT1			•	•	•
9p24	DMental RetardationT2	9p Deletion	158170		•	
9p24.3	DMRT2			•	•	•
9p13.2	Centromere			•	•	•
9q21.11	Centromere			•	•	•
9q22.32	PTCH1	Holoprosencephaly 7	610828	•	•	•
9q22.33	TGFBR1	Loeys-Dietz	609192	•	•	•
9q33.3	NR5A1	XY Sex Reversal	184757	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREGNATAL
9q33.3	LMX1B			•	•	•
9q34.13	TSC1			•	•	•
9q34.3	EHMT1	9q34.3 Microdeletion	610253	•	•	•
9q34.3	Telomere			•	•	•
10p14	GATA3	diGeorge 2	601362	•	•	•
10p12.31	NEBL			•	•	•
10p11.1	Centromere			•	•	•
10q11.21	Centromere			•	•	•
10q11.21	RET	Hirschsprung disease plus, HSCR1	142623	•	•	•
10q21.3	EGR2	Autism	209850	•	•	•
10q23.1	NRG3	10q22-23 Deletion		•	•	•
10q23.2	GRID1	10q22-23 Deletion		•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREGNATAL
10q23.2	BMPR1A	Cowden	158350	•		
10q23.31	PTEN	Mental retardation		•	•	•
10q23.31	SLC16A12	Cataract, juvenile, with microcornea A and glucosuria	612018	•	•	•
10q24.31	LBX1			•	•	•
10q24.32	BTRC			•	•	•
10q24.32	POLL			•	•	•
10q24.32	FBXW4			•	•	•
10q26.11	EMX2	Schizencephaly	269160	•	•	•
10q26.3	Telomere			•	•	•
11p15.5	Telomere			•	•	•
11p15	H19_ICRegion			•	•	•
11p15.5	H19	Beckwith-Wiedemann	130650		•	

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDNATAL
11p15.5	H19_ICregion			•	•	•
11p15.5	IGF2	Beckwith-Wiedemann	130650	•	•	•
11p15.5	INS	Beckwith-Wiedemann	130650	•	•	•
11p15.5	KCNQ1	Long QT	192500	•	•	•
11p15	KCNQ1_ICRegion			•	•	•
11p15.5	KCNQ1_ICregion			•	•	•
11p15.4	CDKN1C	Beckwith-Wiedemann	130650	•	•	•
11p15.4	SLC22A18	Beckwith-Wiedemann	130650	•	•	•
11p15.4	PHLDA2	Beckwith-Wiedemann	130650	•	•	•
11p15.4	OSBPL5			•	•	•
11p15.1	SOX6	Craniosynostosis	128350	•	•	•
11p15.1	ABCC8	Infantile hyperinulinism, enteropathy and deafness	606528 / 276904	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDATAL
11p15.1	USH1C	Infantile hyperinulinism, enteropathy and deafness	606528 / 276904	•	•	•
11p13	PAX6	Aniridia	106210	•	•	•
11p13	WT1			•	•	•
11p11.2	EXT2	Potocki-Shaffer syndrome	601224	•	•	•
11p11.2	ALX4			•	•	•
11p11.2	Centromere			•	•	•
11q11	Centromere			•	•	•
11q13.2	NDUFV1	Leukodystrophy		•	•	•
11q13.4	DHCR7	Smith-Lemli-Opitz syndrome; SLOS	270400	•	•	•
11q14.2	FZD4	11q14 Microdeletion		•	•	•
11q24.3	KCNJ1	Bartter 2	241200	•	•	•
11q25	Telomere			•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREGNATAL
12p13.33	Telomere			•	•	•
12p13.33	CACNA1C	Timothy syndrome; TS	601005	•	•	•
12p11.1	Centromere			•	•	•
12q12	Centromere			•	•	•
12q13.11	COL2A1	Stickler syndrome, type I; STL1	108300	•	•	•
12q14.3	LEMD3			•	•	•
12q14.3	GRIP1	12q14.3 Deletion		•	•	•
12q21.32	CEP290	Joubert 5	610188	•	•	•
12q24.13	PTPN11			•	•	•
12q24.21	TBX5	Holt-Oram	142900	•	•	•
12q24.21	TBX3	Ulnar-mammary syndrome; UMS	181450	•	•	•
12q24.33	Telomere			•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREGNATAL
13q12.11	Centromere			•	•	•
13q12.11	GJB2(Connexin 26)			•	•	•
13q13.1	LGR8(RXFP2)	Cryptorchidism, unilateral or bilateral	219050	•	•	•
13q13.1	BRCA2	Breast cancer	114480	•	•	•
13q14.2	RB1			•	•	•
13q21.32	PCDH9			•	•	•
13q22.3	EDNRB	Hirschsprung disease plus, HSCR2	600155	•	•	•
13q31.3	GPC5	Brachydactyly		•	•	•
13q31.3	GPC6	Brachydactyly		•	•	•
13q32.3	ZIC2	Holopresencaphaly 5	609637	•	•	•
13q34	Telomere			•	•	•
14q11.2	Centromere			•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDNATAL
14q11	MicroDeletionRegion	2p15-p16.1 Microdeletion		•	•	•
14q11.2	SUPT16H			•	•	•
14q11.2	Microdel. Region			•	•	•
14q11.2	CHD8			•	•	•
14q12	FOXP1B	Rett syndrome, congenital variant	613454	•	•	•
14q13.3	PAX9	Tooth agenesis, selective 3; STHAG3	604625	•	•	•
14q22.2	BMP4	14q22 Microdeletion, Orofacial Cleft 11		•	•	•
14q22.3	OTX2	14q22 Microdeletion, microphthalmia, syndromic 5		•	•	•
14q23.1	SIX6	14q22 Microdeletion		•	•	•
14q23.1	SIX1	14q22 Microdeletion		•	•	•
14q23.1	SIX4	14q22 Microdeletion		•	•	•
14q32	DLK1&MEG3_ICRegion			•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREGNATAL
14q32.2	DLK1			•	•	•
14q32.2	DLK1&MEG3_ICregion			•	•	•
14q32.2	MEG3			•	•	•
14q32.33	Telomere			•	•	•
15q11	PWS_ICRegion	Angelman / Prader Willi	105830/176270	•	•	•
15q11	PWS_ICRegion	Angelman / Prader Willi	105830/176270		•	
15q11.2	Centromere				•	
15q11.2	NIPA2	Angelman / Prader Willi	105830/176270	•	•	•
15q11.2	NIPA1	Angelman / Prader Willi	105830/176270	•	•	•
15q11.2	MKRN3	Angelman / Prader Willi	105830/176270	•	•	•
15q11.2	MAGEL2	Angelman / Prader Willi	105830/176270	•	•	•
15q11.2	NDN	Angelman / Prader Willi	105830/176270	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PRENATAL
15q11.2	SNRPN	Angelman / Prader Willi	105830/176270	•	•	•
15q11.2	PWS_ICregion	Angelman / Prader Willi	105830/176270	•	•	•
15q11.2	SNURF	Angelman / Prader Willi	105830/176270	•	•	•
15q11.2	PWS_ICregion	Angelman / Prader Willi	105830/176270	•	•	•
15q11.2	SNORD107/64/108	Angelman / Prader Willi	105830/176270	•	•	•
15q11.2	SnoRNA	Angelman / Prader Willi	105830/176270	•	•	•
15q11.2	SNORD109B	Angelman / Prader Willi	105830/176270	•	•	•
15q11.2	UBE3A			•	•	•
15q12	ATP10A	Autism	209850	•	•	•
15q12	GABRB3	Autism	209850	•	•	•
15q13.1	OCA2	Albinism, oculocutaneous, type II; OCA2	203200	•	•	•
15q13.2	CHRNA7	15q13.3 Microdeletion	612001	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PRENATAL
15q15.3	KIAA0377 (HISPPD2A)	15q15.3 Infertility and deafness		•	•	•
15q15.3	KIAA0377(HISPPD2A)	Infertility and deafness	611102	•	•	•
15q15.3	CATSPER2	Infertility and deafness	611102	•	•	•
15q21.1	SLC12A1	Bartter 1	601678	•	•	•
15q21.1	FBN1	Marfan syndrome; MFS	154700	•	•	•
15q21.2	DMXL2			•	•	•
15q26.1	CHD2	Congenital diaphragmatic hernia	142340	•	•	•
15q26.2	NR2F2	Congenital diaphragmatic hernia	142340	•	•	•
15q26.3	IGF1R	IGF-1 resistance	147370	•	•	•
15q26.3	Telomere			•	•	•
16p13.3	Telomere			•	•	•
16p13	HBA1&HBA2	Alpha thalassemia mental retardation	141750	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDNATAL
16p13.3	HBA1/2			•	•	•
16p13.3	SOX8			•	•	•
16p13.3	TSC2			•	•	•
16p13.3	PKD1	Polycystic kidney disease 1; PKD1	173900	•	•	•
16p13.3	DNASE1	Systemic Lupus erythematosus; SLE	152700	•	•	•
16p13.3	CREBBP			•	•	•
16p13.3	A2BP1			•	•	•
16p11.2	SLC6A8	Creatine deficiency / X-linked mental retardation	300352	•	•	•
16p11.1	Centromere			•	•	•
16q11.2	Centromere			•	•	•
16q12.1	SALL1			•	•	•
16q13	SLC12A3	Gitelman	263800	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREGNATAL
16q21	GPR56	Bilateral frontoparietal polymicrogyria	606854	•	•	•
16q22.1	CBFB	Delayed cranial ossification		•	•	•
16q24.3	FANCA	Fanconi anemia	227650	•	•	•
16q24.3	Telomere			•	•	•
17p13.3	Telomere			•	•	•
17p13.3	ASPA	Canavan	271900	•	•	•
17p13.3	ABR	Medulloblastoma; MDB	155255	•	•	•
17p13.3	YWHAE			•	•	•
17p13.3	PITPNA			•	•	•
17p13.3	PAFAH1B1(LIS1)			•	•	•
17p13.2	CTNS	Cystinosis	219800	•	•	•
17p13.1	TP53	Li-Fraumeni 1	151623	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDAT
17p12	PMP22	Charcot marie tooth disease	118220	•	•	•
17p11.2	RAI1			•	•	•
17p11.2	Centromere			•	•	•
17q11.1	Centromere			•	•	•
17q11.2	SLC6A4	Anxiety, obsessive compulsive disorder; OCD	607834	•	•	•
17q11.2	NF1			•	•	•
17q11.2	JJAZ1(SUZ12)			•	•	•
17q12	TCF2(HNF1B)			•	•	•
17q21.31	SOST	Sclerosteosis 1; SOST1	269500	•	•	•
17q21.31	CRHR1	17q21.31 Microdeletion	610443	•	•	•
17q21.31	MAPT	17q21.31 Microduplication	610443	•	•	•
17q21.32	ITGB3	Glanzmann thrombasthenia; GT	273800	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDNATAL
17q21.33	COL1A1	Osteogenesis imperfecta, type I	166200	•	•	•
17q24.3	SOX9	Campomelic dysplasia	114290	•	•	•
17q25.3	Telomere			•	•	•
18p11.32	Telomere			•	•	•
18p11.31	TGIF1	Holopresencaphaly 4	142946	•	•	•
18p11.21	Centromere			•	•	•
18q11.1	Centromere			•	•	•
18q21.1	TCEB3C			•	•	•
18q21.1	DYM	Dyggve-delchior-clausen disease	223800	•	•	•
18q21.2	MADH4(SMAD4)	Hereditary hemorrhagic telangiectasia	175050	•	•	•
18q21.2	TCF4	Pitt-Hopkins syndrome; PTHS	610954	•	•	•
18q22.3	ZNF407	18q Deletion	301808	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREGNATAL
18q23	GALR1			•	•	•
18q23	Telomere			•	•	•
19p13.3	Telomere			•	•	•
19p13.12	NOTCH3	CADASIL	125310	•	•	•
19p12	Centromere			•	•	•
19q11	Centromere			•	•	•
19q13.2	RPS19	Diamond blackfan anemia	105650	•	•	•
19q13.43	ZIM2			•	•	•
19q13.43	PEG3			•	•	•
19q13.43	PEG3_ICregion			•	•	•
19q13.43	ZNF264			•	•	•
19q13.43	Telomere			•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PRENATAL
20p13	Telomere			•	•	•
20p12.2	JAG1	Alagille	118450	•	•	•
20p11.1	Centromere			•	•	•
20q11.21	Centromere			•	•	•
20q11.22	GDF5	Brachydactyly type C	113100	•	•	•
20q11.23	NNAT			•	•	•
20q13.12	L3MBTL			•	•	•
20q13.2	SALL4	DRRS Duane-Radial Ray syndrome; DRRS	607323	•	•	•
20q13	GNAS_ICRegion			•	•	•
20q13.32	GNAS_ICregion			•	•	•
20q13.32	GNAS	Albright hereditary osteodystrophy	103580	•	•	•
20q13.32	GNAS_ICregion			•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDNATAL
20q13.32	GNAS_ICregion			•	•	•
20q13.33	Telomere			•	•	•
21q11.2	Centromere			•	•	•
21q21.3	APP	Cerebral amyloid angiopathy	605714	•	•	•
21q22.12	DSCR1(RCAN1)	Downs syndrome critical region	190685	•	•	•
21q22.12	RUNX1	Downs syndrome critical region	190685	•	•	•
21q22.13	DSCR3	Downs syndrome critical region	190685	•	•	•
21q22.13	DYRK1A	Downs syndrome critical region	190685	•	•	•
21q22.3	TMEM1	Holoprosencephaly 1	236100	•	•	•
21q22.3	LSS	Holoprosencephaly 1	236100	•	•	•
21q22.3	Telomere			•	•	•
22q11.1	Centromere			•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREGNATAL
22q11	HIRA	diGeorge 1	188400	•	•	•
22q11.21	ATP6V1E1			•	•	•
22q11.21	HIRA			•	•	•
22q11.21	TBX1	diGeorge 1	188400	•	•	•
22q11.21	CRKL	diGeorge 1	188400	•	•	•
22q11.23	BCR	22q11.2 Distal deletion	611867	•	•	•
22q12.2	NF2			•	•	•
22q13.33	ARSA			•	•	•
22q13.33	Telomere			•	•	•
22q13.33	SHANK3	22q13 Microdeletion (Phelan McDermid)	606232	•	•	•
Xp22.33	PAR1			•	•	•
Xp22.33	Telomere			•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDAT
Xp22.33	SHOX			•	•	•
Xp22.33	CDPX1(ARSE)	Chondrodysplasia punctata, X-linked recessive	302950	•	•	•
Xp22.31	NLGN4X	Autism	209850	•	•	•
Xp22.31	VCX3A	X-linked mental retardation		•	•	•
Xp22.31	STS			•	•	•
Xp22.31	VCX3A	X-linked mental retardation		•	•	•
Xp22.31	KAL1	Kallmann 1	308700	•	•	•
Xp22.2	MID1	Opitz GBBB syndrome, X-linked	300000	•	•	•
Xp22.2	HCCS	X-linked mental retardation		•	•	•
Xp22.2	OFD1	Orofacialdigital syndrome I; OFD1	311200	•	•	•
Xp22.2	FANCB	X-linked mental retardation		•	•	•
Xp22.2	AP1S2	X-linked mental retardation		•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDNATAL
Xp22.13	NHS	Nance-Horan syndrome; NHS	302350	•	•	•
Xp22.13	CDKL5	X-linked spasms	300672	•	•	•
Xp22.13	RS1	X-linked juvenile retinoschisis	312700	•	•	•
Xp22.12	PDHA1	Pyruvate dehydrogenase E1-alpha deficiency; PDHAD	312170	•	•	•
Xp22.12	RPS6KA3	Coffin-lowry	303600	•	•	•
Xp22.11	SMS	X-linked mental retardation		•	•	•
Xp22.11	PHEX	Hypophosphatemic rickets	307800	•	•	•
Xp21.3	ARX	X-linked mental retardation		•	•	•
Xp21.3	IL1RAPL1	X-linked mental retardation		•	•	•
Xp21.2	NR0B1(DAX1)	Adrenal hypoplasia congenita	300200	•	•	•
Xp21.2	GK	Glycerol kinase deficiency	307030	•	•	•
Xp21.1	DMD	DMD	310200	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREGNATAL
Xp11.4	CYBB	Chronic granulomatous disease	306400	•	•	•
Xp11.4	OTC	Ornithine transcarbamylase deficiency, hyperammonemia due to	311250	•	•	•
Xp11.4	TM4SF2(TSPAN7)	X-linked mental retardation		•	•	•
Xp11.4	BCOR	X-linked mental retardation		•	•	•
Xp11.4	ATP6AP2	X-linked mental retardation		•	•	•
Xp11.4	CASK	X-linked mental retardation		•	•	•
Xp11.3	MAOA	Brunner	300615	•	•	•
Xp11.3	NDP	Norrie disease; ND	310600	•	•	•
Xp11.23	ZNF674	Xp11.3 deletion	300578	•	•	•
Xp11.23	RP2	Xp11.3 deletion	300578	•	•	•
Xp11.23	ZNF41	X-linked mental retardation		•	•	•
Xp11.23	SYN1	X-linked mental retardation		•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREGNATAL
Xp11.23	ELK1	X-linked mental retardation		•	•	•
Xp11.23	ZNF81	X-linked mental retardation		•	•	•
Xp11.23	SLC38A5	X-linked mental retardation		•	•	•
Xp11.23	FTSJ1	X-linked mental retardation		•	•	•
Xp11.23	PORCN	X-linked mental retardation		•	•	•
Xp11.23	PQBP1	X-linked mental retardation		•	•	•
Xp11.23	CLCN5	Nephrolithiasis, X-linked recessive, with renal failure; XRN	310468	•	•	•
Xp11.22	SHROOM4(KIAA1202)	X-linked mental retardation		•	•	•
Xp11.22	JARID1C	X-linked mental retardation		•	•	•
Xp11.22	SMC1L1(SMC1A)	Cornelia de Lange, X-linked	300590	•	•	•
Xp11.22	HADH2(HSD17B10)	X-linked mental retardation		•	•	•
Xp11.22	HUWE1	X-linked mental retardation, turner	300706	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDICTIVE
Xp11.22	PHF8	X-linked mental retardation		•	•	•
Xp11.22	FGD1	Aarskog-Scott	305400	•	•	•
Xp11.21	KLF8	X-linked mental retardation		•	•	•
Xp11.21	Centromere			•	•	•
Xq11.1	Centromere			•	•	•
Xq11.2	ARHGEF9	Heterotaxy		•	•	•
Xq12	AR	Androgen insensitivity	300068	•	•	•
Xq12	OPHN1	X-linked mental retardation, with cerebellar hypoplasia	300486	•	•	•
Xq13.1	EDA	Ectodermal dysplasia	305100	•	•	•
Xq13.1	DLG3	X-linked mental retardation		•	•	•
Xq13.1	MED12	Opitz-Kaveggia syndrome; OKS	305450	•	•	•
Xq13.1	NLGN3	Autism	209850	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDATAL
Xq13	ZNF261(ZMYM3)	X-linked mental retardation		•	•	•
Xq13.1	ZNF261(ZMYM3)	X-linked mental retardation		•	•	•
Xq13.2	XIST	X inactivation specific transcript		•	•	•
Xq13.2	SLC16A2	X-linked mental retardation, allan-herndon-dudley	300523	•	•	•
Xq13.3	KIAA2022	X-linked mental retardation		•	•	•
Xq13.3	ZDHHC15	X-linked mental retardation		•	•	•
Xq21.1	ATRX	Alpha thalassemia mental retardation	301040	•	•	•
Xq21.1	ATP7A	Menkes disease	309400	•	•	•
Xq21.1	PGK1	Phosphoglycerate kinase 1 deficiency	300653	•	•	•
Xq21.1	BRWD3	X-linked mental retardation		•	•	•
Xq21.2	CHM	Choroideremia	303100	•	•	•
Xq22.1	SRPX2	Rolandic epilepsy, mental retardation, and speech dyspraxia, X-linked	300643	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDICTIVE
Xq22.1	TIMM8A	Deafness-dystonis-optic neuropathy	304700	•	•	•
Xq22.1	BTK	Agammaglobulinemia, X-linked	300755	•	•	•
Xq22.1	GLA	Fabry disease	301500	•	•	•
Xq22.1	NXF5	X-linked mental retardation		•	•	•
Xq22.2	PLP1			•	•	•
Xq22.3	PRPS1	Charcot marie tooth disease, X-linked	311070	•	•	•
Xq22.3	MID2			•	•	•
Xq22.3	COL4A5	Alport, X-linked	301050	•	•	•
Xq23	ACSL4	X-linked mental retardation		•	•	•
Xq23	PAK3	X-linked mental retardation		•	•	•
Xq23	DCX	DCX-associated lissencephaly/subcortical band heterotopia	300067	•	•	•
Xq23	AGTR2	X-linked mental retardation		•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDATAL
Xq24	UBE2A	X-linked mental retardation		•	•	•
Xq24	UPF3B	X-linked mental retardation		•	•	•
Xq24	NDUFA1	X-linked mental retardation		•	•	•
Xq24	LAMP2	Danon disease	300257	•	•	•
Xq24	CUL4B	X-linked mental retardation		•	•	•
Xq25	GRIA3	X-linked mental retardation		•	•	•
Xq25	BIRC4(XIAP)	X-linked lymphoproliferative type 2	308240	•	•	•
Xq25	SH2D1A	X-linked lymphoproliferative type 1	308240	•	•	•
Xq25	OCRL	Lowe oculocerebrorenal syndrome; OCRL	309000	•	•	•
Xq26.1	ZDHHC9	X-linked mental retardation		•	•	•
Xq26.2	GPC3			•	•	•
Xq26.2	PHF6	Borjeson-forssman-lehmann	301900	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDAT
Xq26.2	HPRT1	Lesch-Nyhan	300322	•	•	•
Xq26.3	SLC9A6	X-linked mental retardation, christianson type	300243	•	•	•
Xq26.3	ARHGEF6	X-linked mental retardation		•	•	•
Xq26.3	ZIC3	X-linked heterotaxy	306955	•	•	•
Xq27.1	F9	Hemophilia B	306900	•	•	•
Xq27.1	SOX3	X-linked mental retardation		•	•	•
Xq27.3	FMR1	Fragile X	300624	•	•	•
Xq28	FMR2(AFF2)	Fragile X	300624	•	•	•
Xq28	IDS	Hunter, Mucopolysaccharidosis type 2	309900	•	•	•
Xq28	MTM1	Myopathy, centronuclear, X-linked; CNMX	310400	•	•	•
Xq28	SLC6A8	Creatine deficiency / X-linked mental retardation	300352	•	•	•
Xq28	ABCD1	Adrenoleukodystrophy	300475	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREDATAL
Xq28	L1CAM	HSAS, MASA, CRASH	307000, 303350	•	•	•
Xq28	AVPR2	Diabetes insipidus, nephrogenic, X-linked	304800	•	•	•
Xq28	MECP2			•	•	•
Xq28	FLNA	Heterotopia, periventricular, X-linked dominant	300049	•	•	•
Xq28	RPL10	X-linked mental retardation		•	•	•
Xq28	GDI1	X-linked mental retardation		•	•	•
Xq28	IKBKG	Incontinentia pigmenti	308300	•	•	•
Xq28	DKC1	X-linked dyskeratosis congenita	305000	•	•	•
Xq28	F8	Hemophilia A	306700	•	•	•
Xq28	PAR2			•	•	•
Xq28	Telomere			•	•	•
Yp11.32	Telomere			•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PRENATAL
Yp11.31	SRY			•	•	•
Yp11.2	Centromere			•	•	•
Yq11.21	AZFa	AZFa region	415000	•	•	•
Yq11.21	Centromere			•	•	•
Yq11.223	BPY2	AZFa region	415000	•	•	•
Yq11.223	AZFc	AZFa region	415000	•	•	•
Yq11.223	DAZ1	AZFa region	415000	•	•	•
Yq11.23	CDY1	AZFa region	415000	•	•	•
Yq11.23	GOLGA2LY(AF332229)	AZFa region	415000	•	•	•
Yq11.223	BPY2	AZFa region	415000	•	•	•
Yq11.223	BPY2	AZFa region	415000	•	•	•
Yq11.23	GOLGA2LY(AF332229)	AZFa region	415000	•	•	•

BAND	REGION	DISEASE	OMIM	CYTOARRAY	CYTOARRAY PLUS	CYTOARRAY PREGNATAL
Yq11.23	CDY1	AZFa region	415000	•	•	•
Yq12	Telomere			•	•	•

1.4. CYTOARRAY UPD

The combination of CGH and SNP microarray is a tool that allows not only identifying gains or losses in DNA, but detecting uniparental disomy.

Therefore Cytoarray UPD has identical performance to Cytoarray (analysis of 500 regions with high resolution) and also detects heterozygosity with a resolution of 5Mb. It is therefore an ideal tool when suspicion of uniparental disomy.



1.5. CHROMOSOME ABNORMALITIES DETECTION IN ABORTION REMAINS (DACrA)

Approximately 15% of pregnancies end up in spontaneous abortion. It is estimated that around 1% of couples, have recurrent miscarriages i.e. three or more consecutive losses during 24 months. The cause of the recurrent abortions can vary (age, uterine pathology, hereditary thrombophilia and even nutritional or environmental factors).

However, genetic factors appear to play the most important role. In many cases, one of the members of the couple has a chromosomal abnormality, of which reciprocal translocations are among the most frequent. Reciprocal translocation carriers don't present a particular phenotype, however a large number of gametes with unbalanced translocations are produced resulting miscarriage.

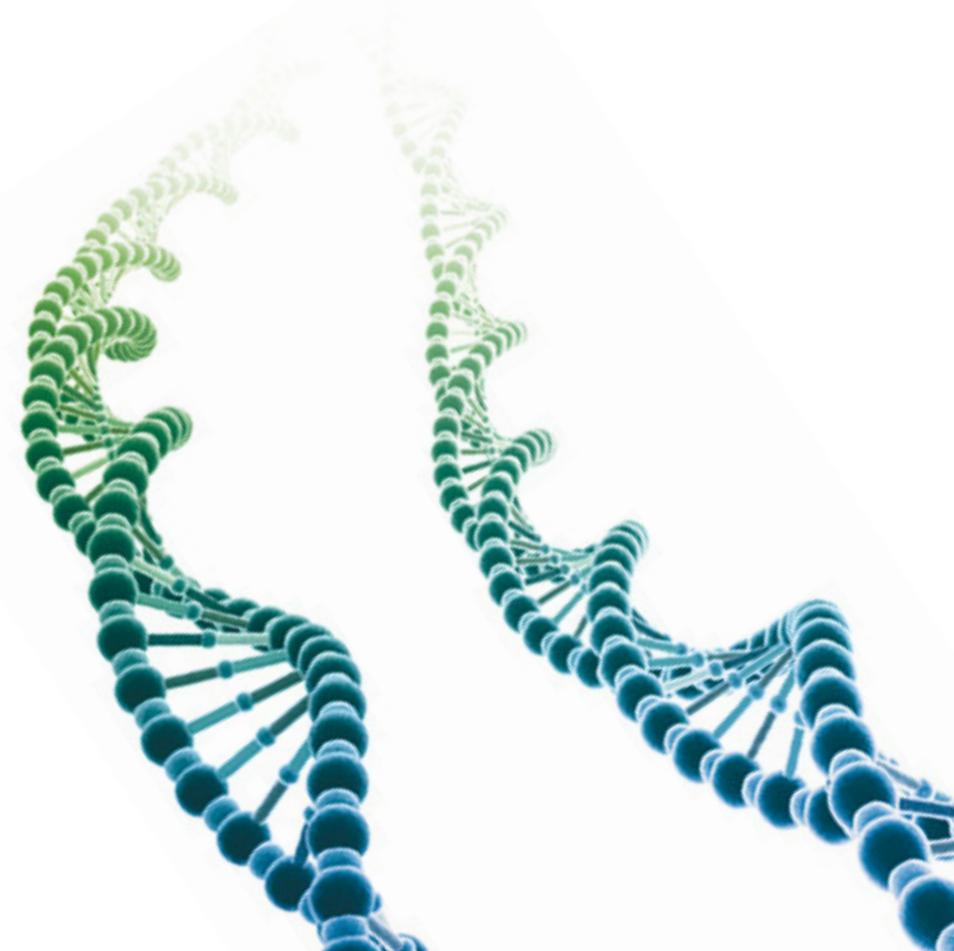
This condition is often analysed by techniques such as karyotype and/or FISH. However, both techniques have different limitations (cell culture requirement, selective growth of maternal cells, test specificity etc.). The DACrA array has been specifically designed to identify copy number alterations from abortion remain tissue, multiplying by 10 the resolution of the karyotype and avoiding the need for culturing cells.

APPLICATIONS

- Unknown cause of recurrent miscarriages.
- Genetic diagnosis in abortions
- Genetic counselling for couples in interrupted pregnancies.
- Aneuploid cells detection.

ADVANTAGES

- Small quantities of starting material required.
- High resolution.
- Cell culture not required.
- Quick results yield.



1.6. HIGH DENSITY CGH MICROARRAY/ 400K and 1M

These are two high-density arrays whose probes are distributed homogeneously along the genome. The 400K format provides an average density of one probe every 5.3 Kb (4.6 Kb in RefSeq genes).

Meanwhile, 1M format provides one probe every 2.1 Kb (1.8 Kb in RefSeq genes). These are more accurate and with more resolution microarrays.

They are both recommended in cases where it is of vital importance accurate determination of the break points, and especially in research.





Parque Científico y Empresarial de la UMH
Edificio Quorum III
03202 · Elche · Alicante · Spain

T: +34 966 682 500
F: +34 966 682 501

info@bioarray.es
www.bioarray.es