



B I O A R R A Y  
Diagnóstico Genético



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|  |           |
|--|-----------|
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MICROARRAY  
CGH

## 1. MICROARRAY CGH

### 1.1. CYTOARRAY

Es el microarray de hibridación genómica comparativa de alta resolución, diseñado específicamente para genética clínica y citogenética.

Detecta variaciones de número de copia de ADN, en pacientes con retraso mental idiopático, autismo y otras alteraciones congénitas. Permite definir si el paciente sufre una enfermedad o síndrome derivados de la presencia de microdelecciones o microduplicaciones en las regiones definidas (ver listado de síndromes que se adjunta).

Basado en el diseño realizado por ISCA (The International Standards for Cytogenomic Arrays Consortium), referente internacional de laboratorios de citogenética, se ha convertido en una herramienta que permite identificar alteraciones de número copia con gran resolución, y una cobertura que permite identificar alteraciones inferiores a 48 Kb en aproximadamente 500 zonas diana.

Cytoarray, con sus más de sesenta mil sondas, ha sido especialmente desarrollado para dar cobertura a numerosos síndromes citogenéticos mediante detección precisa de microdelecciones y microduplicaciones, superando así a las técnicas citogenéticas convencionales.

#### CYTOARRAY PERMITE DETECTAR

- Aneuploidias
- Alteraciones de alrededor de 48 Kb en cerca de 500 zonas diana.
- Más de 400 síndromes (una sonda cada 31kb)
- 233 genes asociados con autismo, retraso mental, cardiopatías congénitas y enfermedades oculares.
- Bandas subteloméricas (una sonda cada 75 kb)

En definitiva, se trata de una potente herramienta diagnóstica que permite obtener mayor información que los métodos tradicionales para un diagnóstico más efectivo.

## 1.2. CYTOARRAY PLUS

Con un diseño similar a Cytoarray, pero con una mayor definición y resolución, Cytoarray Plus con sus ciento ochenta mil sondas, ha sido especialmente desarrollado para identificar alteraciones muy pequeñas en múltiples genes relacionados con patología.

### CARACTERÍSTICAS TÉCNICAS

- Densidad de sonda de una cada 25kb y una resolución de una sonda cada 14kb, aportando una media de 6 sondas por gen.
- 501 regiones de genes asociados a autismo, retraso mental, cardiopatías congénitas y enfermedades oculares.



### 1.3. CYTOARRAY DIAGNÓSTICO PRENATAL

Es un microarray desarrollado específicamente para el diagnóstico prenatal que permite detectar cerca de 500 síndromes asociados a 233 genes. Cytoarray, con sus más de sesenta mil sondas y su amplia cobertura del genoma es una herramienta perfecta para diagnóstico prenatal, permitiendo obtener resultados en menos de una semana.

#### APLICACIONES

- Cariotipos con anomalías que requieren aclaración.
- En los casos de sospecha ecográfica de fetos polimalformados.
- Gestaciones previas con muerte fetal de causa no establecida o pérdida recurrente de la gestación
- Antecedentes de enfermedades genéticas en hijos previos.
- Ansiedad materna.



## TABLA DE SÍNDROMES

| BANDA   | REGIÓN     | PATOLOGÍA                        | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|------------|----------------------------------|--------|-----------|----------------|--------------------|
| 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 |                                  |        | •         | •              | •                  |

| BANDA  | REGIÓN   | PATOLOGÍA  | 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     |  |        | •         | •              | •                  |

| BANDA  | REGIÓN              | PATOLOGÍA                                | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|--------|---------------------|--|--------|-----------|----------------|--------------------|
| 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 | •         | •              | •                  |

| BANDA  | REGIÓN     | PATOLOGÍA   | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|--------|------------|---|--------|-----------|----------------|--------------------|
| 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 pallet  | 119540 | •         | •              | •                  |
| 2q33.1 | BMPR2      | Pulmonary hypertension, primary 1; PPH1,                        | 178600 | •         | •              | •                  |

| BANDA   | REGIÓN     | PATOLOGÍA                                    | 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 |  |        | •         | •              | •                  |

| BANDA   | REGIÓN     | PATOLOGÍA                                    | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|------------|--|--------|-----------|----------------|--------------------|
| 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      |  |        | •         | •              | •                  |

| BANDA   | REGIÓN     | PATOLOGÍA                         | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|------------|-----------------------------------|--------|-----------|----------------|--------------------|
| 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 |                                   |        | •         | •              | •                  |

| BANDA   | REGIÓN       | PATOLOGÍA   | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|--------------|---|--------|-----------|----------------|--------------------|
| 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 | •         | •              | •                  |

| BANDA   | REGIÓN         | PATOLOGÍA  | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|----------------|--|--------|-----------|----------------|--------------------|
| 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       |  |        | •         | •              | •                  |

| BANDA   | REGIÓN     | PATOLOGÍA   | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|------------|---|--------|-----------|----------------|--------------------|
| 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 | •         | •              | •                  |

| BANDA   | REGIÓN         | PATOLOGÍA  | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|----------------|--|--------|-----------|----------------|--------------------|
| 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           |  |        | •         | •              | •                  |

| BANDA  | REGIÓN      | PATOLOGÍA                      | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|--------|-------------|--------------------------------|--------|-----------|----------------|--------------------|
| 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 | •         | •              | •                  |

| BANDA   | REGIÓN      | PATOLOGÍA          | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|-------------|--------------------|--------|-----------|----------------|--------------------|
| 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 | •         | •              | •                  |

| BANDA   | REGIÓN                | PATOLOGÍA   | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|-----------------------|---|--------|-----------|----------------|--------------------|
| 8q24.3  | KCNK9                 | Birk-Barel mentl 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 | •         | •              | •                  |

| BANDA    | REGIÓN     | PATOLOGÍA                        | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|----------|------------|----------------------------------|--------|-----------|----------------|--------------------|
| 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                |        | •         | •              | •                  |

| BANDA    | REGIÓN       | PATOLOGÍA   | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|----------|--------------|---|--------|-----------|----------------|--------------------|
| 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 |           | •              |                    |

| BANDA   | REGIÓN         | PATOLOGÍA  | OMIM            | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|----------------|--|-----------------|-----------|----------------|--------------------|
| 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 | •         | •              | •                  |

| BANDA   | REGIÓN     | PATOLOGÍA  | OMIM            | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|------------|--|-----------------|-----------|----------------|--------------------|
| 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   |  |                 | •         | •              | •                  |

| BANDA    | REGIÓN     | PATOLOGÍA                       | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|----------|------------|---------------------------------|--------|-----------|----------------|--------------------|
| 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   |                                 |        | •         | •              | •                  |

| BANDA    | REGIÓN            | PATOLOGÍA                               | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|----------|-------------------|---|--------|-----------|----------------|--------------------|
| 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              | Holoprosencaphaly 5                     | 609637 | •         | •              | •                  |
| 13q34    | Telomere          |   |        | •         | •              | •                  |
| 14q11.2  | Centromere        |   |        | •         | •              | •                  |

| BANDA   | REGIÓN              | PATOLOGÍA  | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|---------------------|--|--------|-----------|----------------|--------------------|
| 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  |  |        | •         | •              | •                  |

| BANDA    | REGIÓN             | PATOLOGÍA               | OMIM          | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|----------|--------------------|-------------------------|---------------|-----------|----------------|--------------------|
| 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 | •         | •              | •                  |

| BANDA   | REGIÓN          | PATOLOGÍA                               | 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        | •         | •              | •                  |

| BANDA   | REGIÓN              | PATOLOGÍA                            | 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 | •         | •              | •                  |

| BANDA   | REGIÓN     | PATOLOGÍA   | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|------------|---|--------|-----------|----------------|--------------------|
| 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 | •         | •              | •                  |

| BANDA   | REGIÓN         | PATOLOGÍA                               | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|----------------|---|--------|-----------|----------------|--------------------|
| 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 | •         | •              | •                  |

| BANDA    | REGIÓN       | PATOLOGÍA                                   | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|----------|--------------|---|--------|-----------|----------------|--------------------|
| 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 | •         | •              | •                  |

| BANDA    | REGIÓN       | PATOLOGÍA                             | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|----------|--------------|---------------------------------------|--------|-----------|----------------|--------------------|
| 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 | •         | •              | •                  |

| BANDA    | REGIÓN        | PATOLOGÍA               | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|----------|---------------|-------------------------|--------|-----------|----------------|--------------------|
| 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      |                         |        | •         | •              | •                  |

| BANDA    | REGIÓN        | PATOLOGÍA                            | 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 |                                      |        | •         | •              | •                  |

| BANDA    | REGIÓN        | PATOLOGÍA                      | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|----------|---------------|--------------------------------|--------|-----------|----------------|--------------------|
| 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    |                                |        | •         | •              | •                  |

| BANDA    | REGIÓN   | PATOLOGÍA                             | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|----------|----------|---------------------------------------|--------|-----------|----------------|--------------------|
| 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 |                                       |        | •         | •              | •                  |

| BANDA   | REGIÓN      | PATOLOGÍA                                     | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|-------------|---|--------|-----------|----------------|--------------------|
| 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                   |        | •         | •              | •                  |

| BANDA   | REGIÓN      | PATOLOGÍA   | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|-------------|---|--------|-----------|----------------|--------------------|
| 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 | •         | •              | •                  |

| BANDA   | REGIÓN         | PATOLOGÍA  | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|----------------|--|--------|-----------|----------------|--------------------|
| 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                                  |        | •         | •              | •                  |

| BANDA   | REGIÓN            | PATOLOGÍA  | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|-------------------|--|--------|-----------|----------------|--------------------|
| 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 | •         | •              | •                  |

| BANDA   | REGIÓN     | PATOLOGÍA   | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|------------|---|--------|-----------|----------------|--------------------|
| 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 | •         | •              | •                  |

| BANDA  | REGIÓN        | PATOLOGÍA   | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|--------|---------------|---|--------|-----------|----------------|--------------------|
| 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 | •         | •              | •                  |

| BANDA  | REGIÓN | PATOLOGÍA   | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|--------|--------|---|--------|-----------|----------------|--------------------|
| 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                               |        | •         | •              | •                  |

| BANDA  | REGIÓN      | PATOLOGÍA                             | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|--------|-------------|---------------------------------------|--------|-----------|----------------|--------------------|
| 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 | •         | •              | •                  |

| BANDA  | REGIÓN     | PATOLOGÍA   | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|--------|------------|---|--------|-----------|----------------|--------------------|
| 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 | •         | •              | •                  |

| BANDA   | REGIÓN   | PATOLOGÍA                                       | OMIM           | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|----------|---|----------------|-----------|----------------|--------------------|
| 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 |   |                | •         | •              | •                  |

| BANDA    | REGIÓN             | PATOLOGÍA   | 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 | •         | •              | •                  |

| BANDA   | REGIÓN   | PATOLOGÍA   | OMIM   | CYTOARRAY | CYTOARRAY PLUS | CYTOARRAY PRENATAL |
|---------|----------|-------------|--------|-----------|----------------|--------------------|
| Yq11.23 | CDY1     | AZFa region | 415000 | •         | •              | •                  |
| Yq12    | Telomere |             |        | •         | •              | •                  |

## 1.4. CYTOARRAY UPD

La combinación de un microarray CGH y SNPs es una herramienta que permite no sólo identificar ganancias o pérdidas de material genético, si no también detectar disomías uniparentales.

El análisis Cytoarray UPD tiene idénticas prestaciones al Cytoarray (análisis de 500 regiones con gran resolución) y además detecta pérdidas de heterocigosidad con una resolución de 5Mb. Se trata, por tanto, de la herramienta ideal cuando existe sospecha de una patología relacionada con disomías uniparentales.



## 1.5. DETECCIÓN DE ANOMALÍAS CROMOSÓMICAS EN RESTOS ABORTIVOS (DACrA)

Aproximadamente el 15% de los embarazos acaban en aborto espontáneo. En el 1% de las parejas, además, estos abortos se producen de manera recurrente, es decir se producen tres o más pérdidas consecutivas antes de los 24 meses. La causa de los abortos de repetición puede ser variada (edad, patología uterina, trombofilia hereditaria e incluso factores nutricionales y ambientales).

Sin embargo, los factores genéticos parecen jugar el papel más importante. En muchas ocasiones, alguno de los miembros de la pareja tiene una anomalía cromosómica, siendo las translocaciones recíprocas una de las más frecuentes. Los portadores de translocaciones recíprocas no presentan un fenotipo particular, pero las parejas sufren de abortos espontáneos porque producen un gran número de gametos con translocaciones no balanceadas.

Es muy común que los estudios citogenéticos de abortos de repetición se realicen mediante cariotipo y/o FISH. Sin embargo, ambas técnicas tienen diferentes limitaciones (requiere cultivo de células, crecimiento selectivo de células maternas, especificada de la prueba, etc.). El array DACrA ha sido específicamente diseñado para identificar alteraciones de número de copia en restos abortivos, multiplicando por 10 la resolución del cariotipo y evitando la necesidad de cultivar células.

### APLICACIONES

- Abortos de repetición de causa desconocida.
- Diagnóstico genético en abortos
- Consejo genético en parejas con embarazos interrumpidos.
- Detección de aneuploidías.

## VENTAJAS

- Poco material de partida.
- Mayor resolución.
- No es necesario cultivar células.
- Rápidez en el diagnóstico.



## 1.6. MICROARRAY CGH ALTA DENSIDAD 400K Y 1M

Se trata de dos arrays de alta densidad cuyas sondas están distribuidas de forma homogénea a lo largo de todo el genoma. El formato 400K proporciona una cobertura con una densidad media de una sonda cada 5,3kb (4,6kb en genes RefSeq). Por su parte, el formato 1M dispone una sonda cada 2,1kb (1,8Kb en genes RefSeq). Se trata por tanto de los microarrays con mayor precisión y resolución.

Su aplicación está recomendada en casos donde es de vital importancia la determinación precisa de los puntos de rotura, y especialmente en investigación.





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