INDICATIVE DETECTION RATES OF KNOWN GENETIC DISEASES AND SYNDROMES THROUGH PRENATAL 2x105K MICROARRAY

A. HIGH DETECTION RATES [percent detection ranges from 50 to over 99%]

DISORDER/SYNDROME	CHROMOSOME REGION
1p32-p31 deletion syndrome	1p32-p31
1p36 deletion and/or duplication/triplication	1p36
1q21.1 deletion syndrome	1q21.1
1q21.1 duplication syndrome	1q21.1
1q41-q42 deletion syndrome	1q41-q42
1q43-q44 deletion syndrome	1q43-q44
2p12-p11.2 deletion syndrome	2p12-p11.2
2p16.1-p15 deletion syndrome	2p16.1-p15
2q31.1 duplication syndrome	2q31.1
3q13.31 deletion syndrome	3q13.31
3q29 microdeletion syndrome	3q29
3q29 microduplication syndrome	3q29
4q21 deletion syndrome	4q21
5p13 microduplication syndrome	5p13
5q12 deletion syndrome	5q12

5q14.3 deletion syndrome	5q14.3
6pter-p24 deletion syndrome	6pter-p24
6q11-q14 deletion syndrome	6q11-q14
6q25-q25 deletion syndrome	6q24-q25
7q11.23 deletion syndrome, distal, 1.2Mb	7q11.23
8p inversion duplication/deletion	8p23.1
8q21.11 deletion syndrome	8q21.11
9p24.3 deletion syndrome/gonadal dysgenesis	9p24.3
9q34.3 deletion syndrome	9q34.3
10q23 deletion syndrome	10q23
10q26 deletion syndrome	10q26
11p15-p14 **homozygous** deletion syndrome	11p15-p14
14q11q22 deletion syndrome	14q11-q22
15q11.2q12(PWS AS) duplication	15q11.2-q12
15q13.3 microdeletion syndrome	15q13.3
15q21 deletion syndrome	15q21
15q24 deletion syndrome	15q24
15q25 deletion syndrome	15q25
15q26-qter deletion syndrome	15q26-qter
16p12.1 deletion syndrome, 520kb	16p12

16p12.2p11.2 deletion syndrome	16p12.2-p11.2
16q22 deletion syndrome	16q22
17p13.1 deletion syndrome	17p13.1
17q11.2 deletion syndrome, 1.4Mb	17q11.2
17q12 deletion syndrome (RCAD)	17q12
17q12 duplication syndrome	17q12
17q21.31 duplication syndrome	17q21.31
17q23.1-q23.2 deletion syndrome	17q23.1-q23.2
18p deletion syndrome	18p
18q deletion syndrome	18q
19p13.13 deletion syndrome	19p13.13
19q13.11 deletion syndrome	19q13.11
22q11.2 duplication syndrome	22q11.2
Xp11.23-p11.22 duplication syndrome	Xp11.23-p11.22
Xp11.3 deletion syndrome	Xp11.3
Xp21 deletion syndrome	Xp21
Xq27.3q28 duplication syndrome	Xq27.3q28
Angelman syndrome	15q11.2-q12 deletion
Cat eye syndrome	inv dup(22)(q11.2)

Congenital adrenal hypoplasia	Xp21.2 deletion
Cri-du-Chat syndrome	5p15.2p13.3 deletion
DiGeorge, Velocardiofacial syndrome	22q11.2 deletion
DiGeorge syndrome 2	10p14 deletion
Dosage sensitive sex reversal	Xp21.2 duplication
Jacobsen (11q25 deletion) syndrome	11q24-q25
Langer-Giedion syndrome	8q23.3q24.11 deletion
Leri-Weill dyschondrosteosis	Xp22.33/Yp11.32
Miller-Dieker lissencephaly syndrome	17p13.3 deletion
Nephronophthisis 1, due to **homozygous** NPH1 deletion	2q13 **homozygous** deletion
Pelizaeus-Merzbacher disease	Xq22 duplication or deletion
Prader-Willi syndrome	15q11.2q12 deletion
Prader-Willi/Angelman duplication	15q11.2q12
Potocki-Shaffer syndrome	11p11.2 deletion
Smith-Magenis syndrome	17p11.2 deletion
Smith-Magenis syndrome duplication	17p11.2 duplication
Split-Hand/foot malformation-5	2q31 deletion
Steroid Sulfatase Deficiency	Xp22.31 deletion/duplication
Williams-Beuren syndrome	7q11.23 deletion/duplication
Wolf-Hirschhorn syndrome	4p16.3 deletion

B. INTERMEDIATE DETECTION RATES [percent detection ranges from 5 to 35%]

DISORDER	CHROMOSOME REGION
2q22.3 deletion syndrome	2q22.3
Alagille syndrome	20p12.2 deletion
Aniridia	11p13 deletion
Duchenne/Becker muscular dystrophy	Xp21.2
Glycerol kinase deficiency	Xp22 deletion
Greig cephalo-polysyndactyly syndrome	7p14.1 deletion
Neurofibromatosis II	22q12.2 deletion
Retinoblastoma	13q14.2 deletion
Rett syndrome	Xq28 deletion/duplication
Rubinstein-Taybi syndrome	16p13.3 deletion
Saethre-Chotzen syndrome	7p21.1 deletion
Sex reversal X/Y translocations	Yp11.31 deletion
Trichorhinophalangeal syndrome 1	8q23.3 deletion
Tuberous sclerosis 1	9q34.2 deletion
WAGR syndrome	11p13 deletion
Wilms tumor	11p13 deletion

C. VERY LOW DETECTION RATES [percent detection less than 5%]

DISORDER	CHROMOSOME REGION
Autism spectrum	15q11.2q13, 16p11.2, 22q13.3, Xp22.32
Basal cell nevus syndrome	9q22.32 deletion
Beckwith-Wiedemann syndrome	11p15.5 deletion/duplication
Bruton agammaglobulinemia	Xq22.1 deletion
Campomelic dysplasia	17q24.3 deletion
CHARGE syndrome	8q12.2 deletion
Cleidocranial dysplasia	6p21.1 deletion
Congenital diaphragmatic hernia	15q26.1q26.2 deletion
Cornelia de Lange syndrome	5p13.2 deletion
Dandy-Walker syndrome	3q24 deletion
Feingold syndrome	2p24.3 deletion
Holoprosencephaly 1	21q22.3 deletion
Holoprosencephaly 2	2p21 deletion
Holoprosencephaly 3	7q36.3 deletion
Holoprosencephaly 4	18p11.31 deletion
Holoprosencephaly 5	13q32.3 deletion
Hypoparathyroidism, sensorineural deafness and renal disease	10p14 duplication
Kallmann syndrome 1	Xp22.31 deletion

Leukodystrophy	11q14.2q14.3 deletion
Mental retardation X-linked with growth hormone deficiency	Xq27.1 deletion or duplication
Microphthalmia with linear skin defects	Xp22.2 deletion
Nail-patella syndrome	9q33.3 deletion
Neurofibromatosis I	17q11.2 deletion
Noonan syndrome	12q24.13 deletion
Rieger syndrome, type 1	4q25 deletion
Split-Hand/foot malformation-3	10p14 duplication
Sotos syndrome	5q35 deletion
X-linked heterotaxy	Xq26.3 deletion

IMPORTANT NOTES: Microdeletion/microduplication analysis through prenatal microarray establishes a diagnosis in the vast majority of disorders listed above under **Section A**.

The occurrence of microdeletions/ microduplications in disorders listed in **Sections B and C** is much less frequent or very infrequent, as these disorders are due predominantly to the presence of gene mutations, not detectable through molecular karyotype-aCGH.

Furthermore, a number of microdeletion/microduplication syndromes may present with variable penetrance and expressivity, meaning that not all persons with the microdeletion/microduplication will be affected or that the clinical symptoms may vary considerably.

In general, reporting will include microdeletions/microduplications and other chromosomal imbalances of known clinical significance. Assessment and reporting of other microdeletions/microduplications will be determined accordingly, following expert clinical evaluation. In selected cases and depending on the findings, parental studies may be required prior to the final report.

May we remind that InterGenetics offers mutation detection, via Next Generation Sequencing - NGS or classic DNA Sanger sequencing, for most all the disorders listed in the Table above, such as Noonan syndrome, Rett syndrome, CHARGE syndrome, Tuberous Sclerosis, etc..