Cat Nr/REF: KBI-40006

English

For professional use only

Poseidon™ Repeat Free™ Chromosome 13/21, X/Y/18 specific DNA Probes

Introduction:

Trisomy 21 is one of the most common chromosomal abnormalities in live born children and causes Down syndrome, a particular combination of phenotypic features that includes mental retardation and characteristic facies. Molecular analysis has revealed that the 21q22.1-q22.3 region appears to contain the gene(s) responsible for the congenital heart disease observed in Down syndrome.

Trisomy 13, also called Patau syndrome, is a chromosomal condition that is associated with severe mental retardation and certain physical abnormalities. The critical region has been reported to include 13q14-13q32 with variable expression, gene interactions, or interchromosomal effects.

Trisomy 18 causing Edwards syndrome is the second most common autosomal trisomy after trisomy 21.

The disorder/condition is characterized by severe psychomotor and growth retardation, microcephaly, microphthalmia, malformed ears, micrognathia or retrognathia, microstomia, distinctively clenched fingers, and other congenital malformations.

Chromosomal abnormalities involving the X and Y chromosome (sex chromosomes) are slightly less common than autosomal abnormalities and are usually much less severe in their effects. The high frequency of people with sex chromosome aberrations is partly due to the fact that they are rarely lethal conditions.

Turner syndrome occurs when females inherit only one X chromosome; their genotype is X0.

Metafemales or triple-X females, inherit three X chromosomes; their genotype is XXX or more rarely XXXX or XXXXX.

Klinefelter syndrome males inherit one or more extra X chromosomes; their genotype is XXY or more rarely XXXY, XXXXY, or XY/XXY mosaic.

XYY syndrome males inherit an extra Y chromosome; their genotype is XYY.

Intended use:

The **chromosome 21 specific** region probe is optimized to detect copy numbers of chromosome 21 at 21q22.1 on uncultured amniotic cells.

The **chromosome 13 specific** region probes is optimized to detect copy numbers of Chromosome 13 at 13q14.2 on uncultured amniotic cells.

The **chromosome 18 specific** Satellite probe (D18Z1) is optimized to detect copy numbers of Chromosome 18 at 18p11-18q11 on uncultured amniotic cells.

The **chromosome X specific** Satellite probe (DXZ1) is optimized to detect copy numbers of Chromosome X at Xp11-Xq11 on uncultured amniotic cells.

The **chromosome Y specific** Satellite probe (DYZ3) is optimized to detect copy numbers of Chromosome Y at Yp11-Yq11 on uncultured amniotic cells.

These probes are recommended to be used in combination with the Poseidon FISH & Digestion Kit (KBI-60003) providing necessary reagents to perform FISH for optimal results.

Vial 1

Critical region 1 (red): The **21q specific** DNA probe is direct-labeled with Platinum*Bright*550.

Critical region 2 (green): The **13q14 specific** DNA probe is direct-labeled with Platinum*Bright*495.

Vial 2

Critical region 3 (blue): The **18 SE** DNA probe is direct-labeled with Platinum*Bright*415.

Critical region 4 (green): The **X SE** DNA probe is direct-labeled with Platinum*Bright*495.

Critical region 5 (red): The **Y SE** DNA probe is direct-labeled with Platinum*Bright*550.

More information on the backside

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POSEIDON™
REPEAT-FREE™ FISH PROBES

Application Manual

KBI-40006

PrenatScreen (13/ 21, X/ Y/18)

IVD
for EU only

CE 0344

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2°C 8°C long term storage

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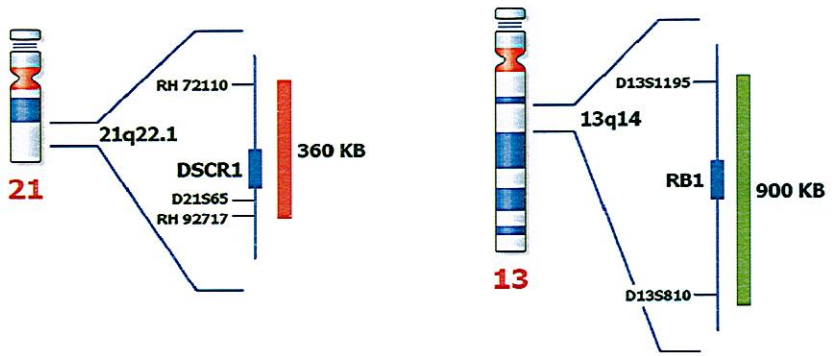

Not to scale

Application manual

	Normal Signal Pattern		Trisomy 21	Trisomy 13	Trisomy 18	
Expected Signals Using 13/21	2R2G		3R2G	2R3G		
Expected Signals Using 13/21+18	2R2G2B		3R2G2B	2R3G2B	2R2G3B	
	Female	Male			Female	Male
Expected Signals Using X/Y + 18	2G2B	1R1G2B			2G3B	1R1G3B
	Female	Male	Turner XO	Meta-female	Klinefelter	XYY
Expected Signals Using X/Y	2G	1R1G	1G	3-5G	2G1R 3-4G1R 1R1G/1R2G in mosaics	1G2R
Expected Signals Using X/Y + 18	2G2B	1R1G2B	1G2B	3-5G2B	2G1R2B 3-4G1R2B 1R1G2B/1R2G2B in mosaics	1G2R2B

Interpretation table:

Reagent:

Poseidon probes are direct-labeled DNA probes provided in a ready-to-use format. Apply 10 µl of probe to a sample area of approximately 22 x 22 mm.

Please refer to the Instructions for Use for the entire Poseidon FISH protocol.

The chromosome 13/21 specific probe is a Poseidon Repeat Free probe which does not contain Cot-1 DNA. Hybridization efficiency is therefore increased and background, due to unspecific binding, is highly reduced.

Interpretation:

The **Chromosome 13/21 specific** probe is designed as a dual-color assay to detect gains of chromosome 21 and 13. Trisomy 21 will be detected by three red signal at the 21q22 region and two green signals for chromosome 13 (3R2G). Trisomy 13 will be detected by 3 green signals at the 13q14 region and two red signals for chromosome 21 (2R3G).
Two single color red (R) and green (G) signals will identify the normal chromosomes 13 and 21 (2R2G). The **Chromosome X/Y/18 specific** probe is designed as a triple-colour assay to detect gains or losses of chromosome X, Y and or 18. Turner syndrome will be detected by one green signal only at Xcen. Meta-Females (or Triple-X females) will be detected by three or more green signals at Xcen. Klinefelter will be detected by 2 or more green and 1 red signal. XYY males will be detected by one green and two red signals. Two single green (G) signals will identify the normal X chromosome in females, one green and one red signal will identify the normal X and Y chromosomes in male. Trisomy 18 will be detected by three blue signals at 18 cen. Two single blue signals will identify the normal chromosome 18.

References:

Korenberg J. et al, 1994, Proc. Nat. Acad. Sci. 91; 4997-5001
Spathas D et al, 1994, Prenat Diagn. 14(11); 1049-1054
Tepperberg et al, 2001, Prenat Diagn 21(4); 293-301